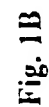


**Fig. 1A**

[illegible]

**Fig. 1B**



**Family FHA - 1**

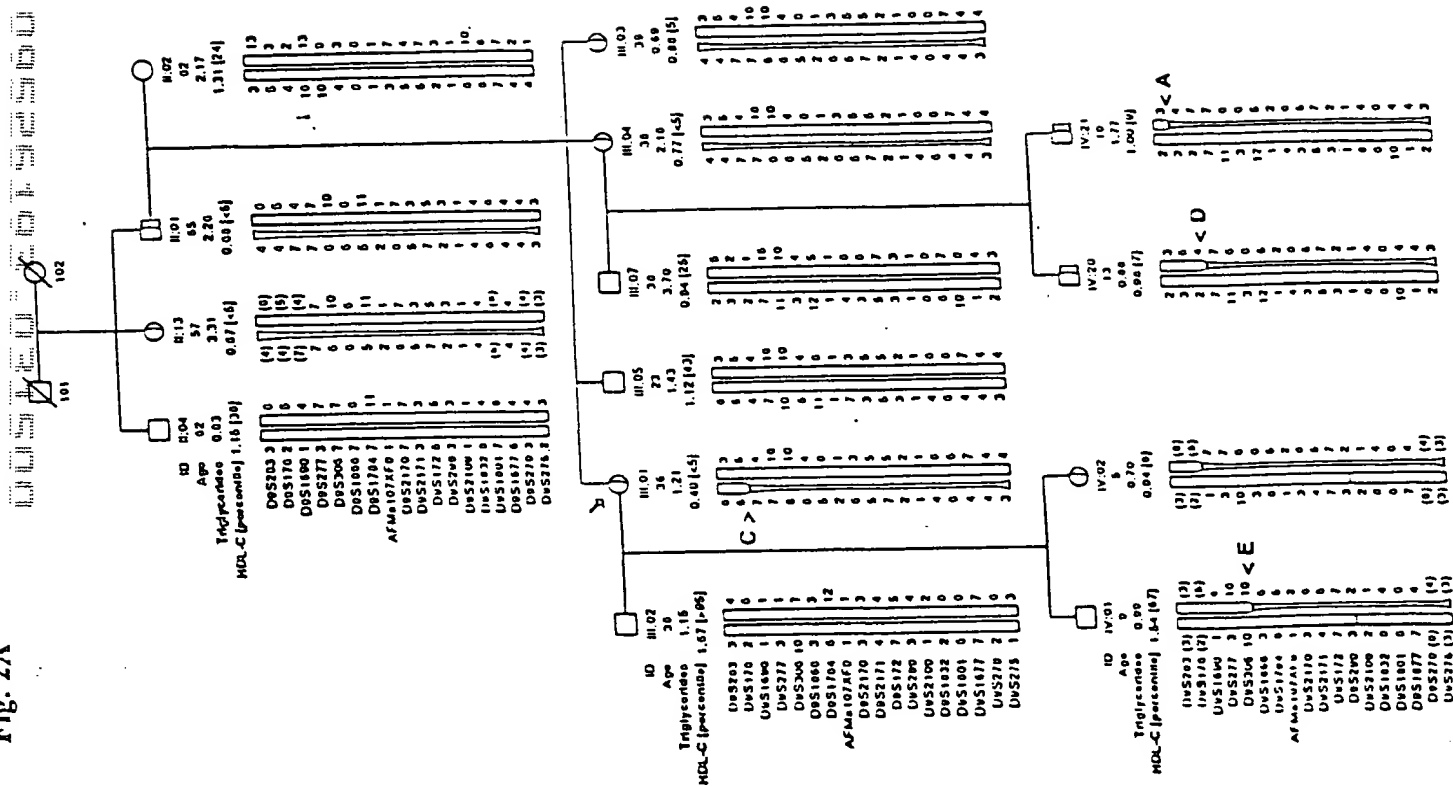
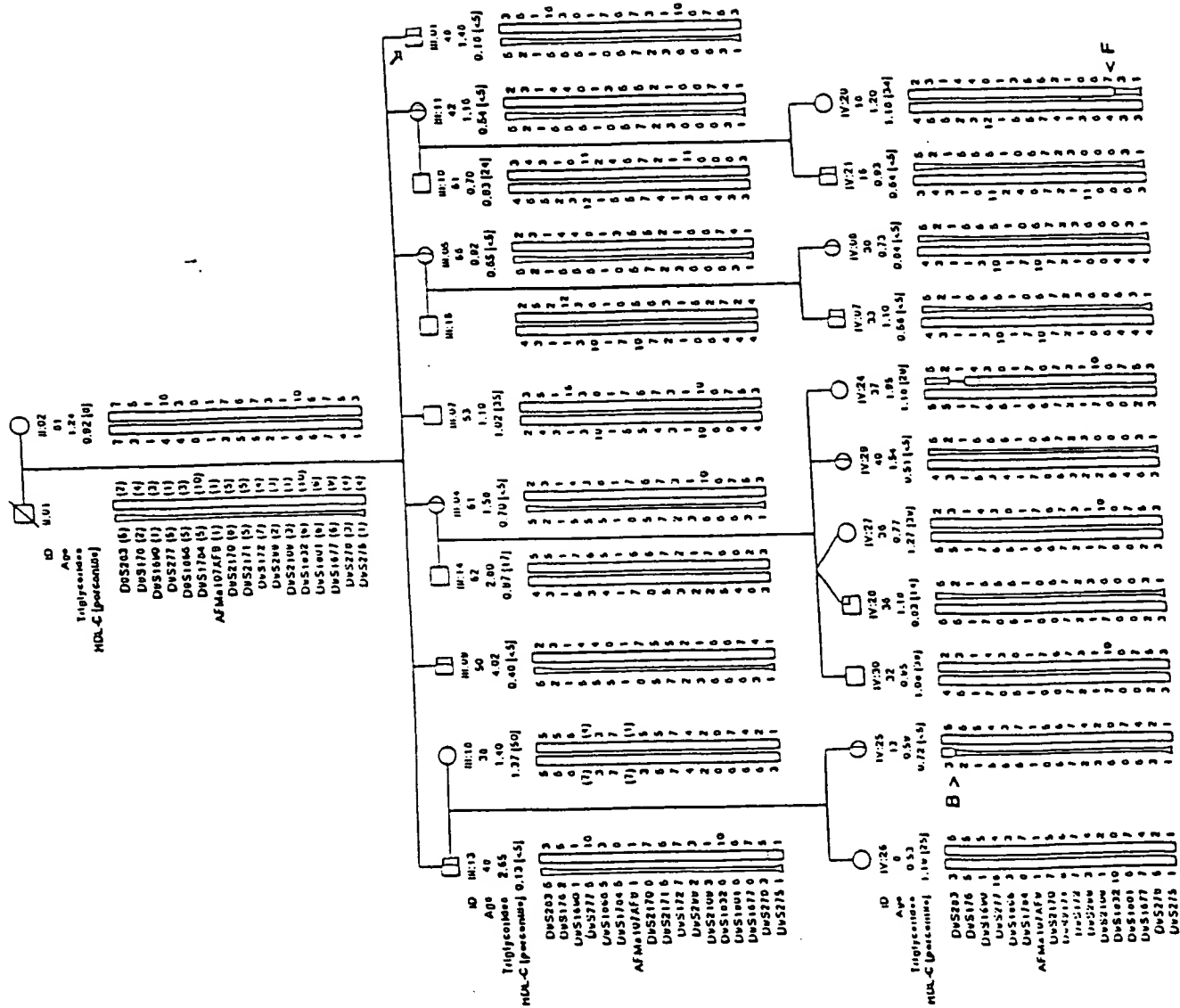


Fig. 2B

Family FHA - 2



Family FHA - 3

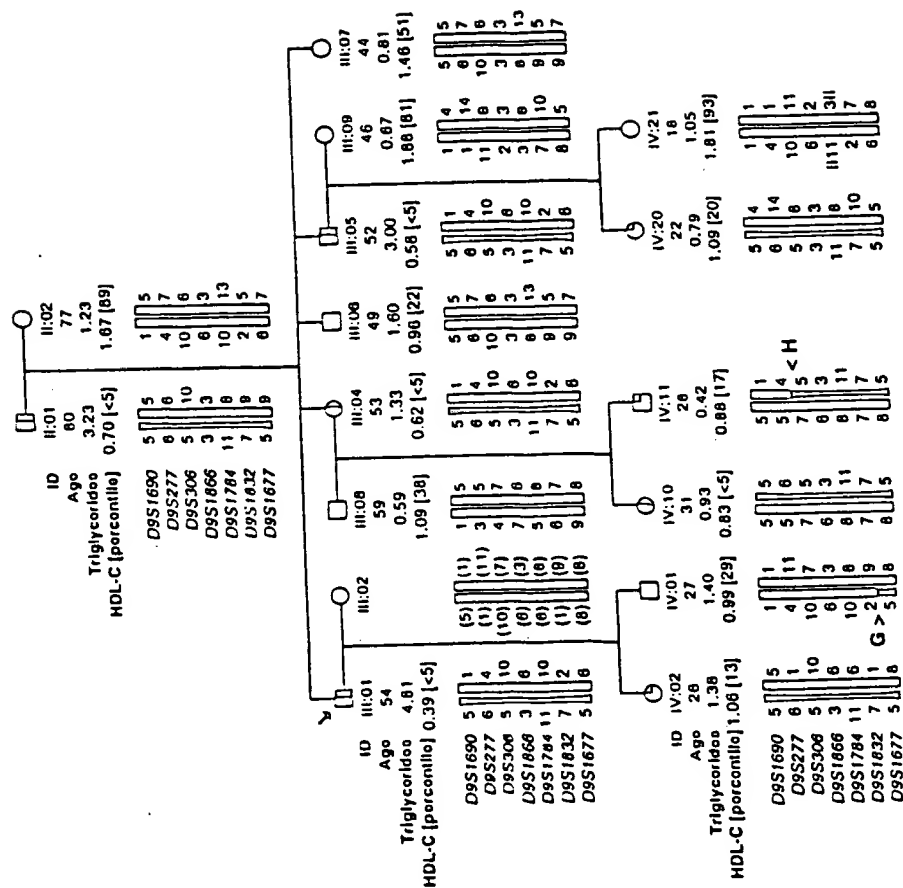
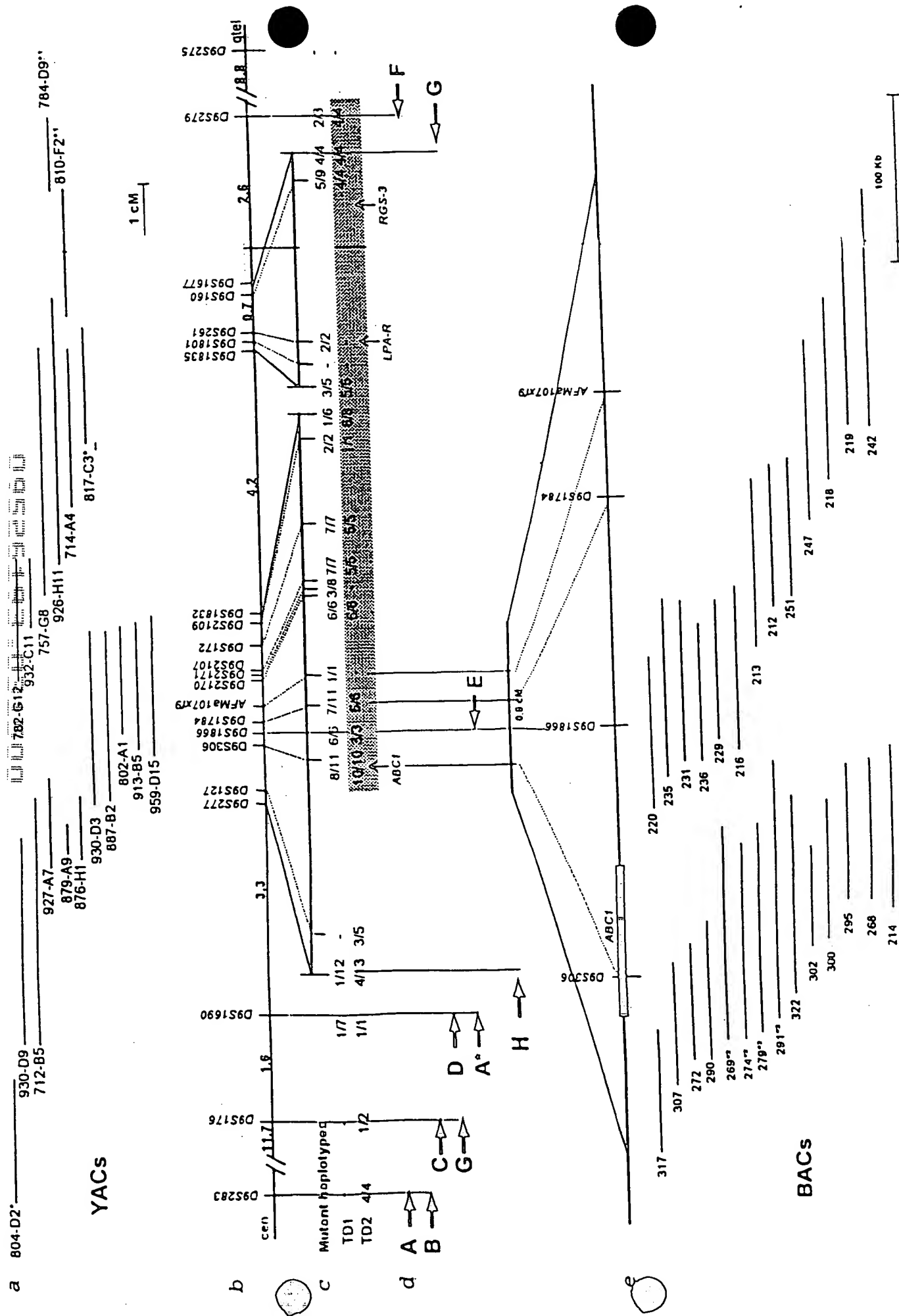


Fig. 2C







# Exon 30 mutation:

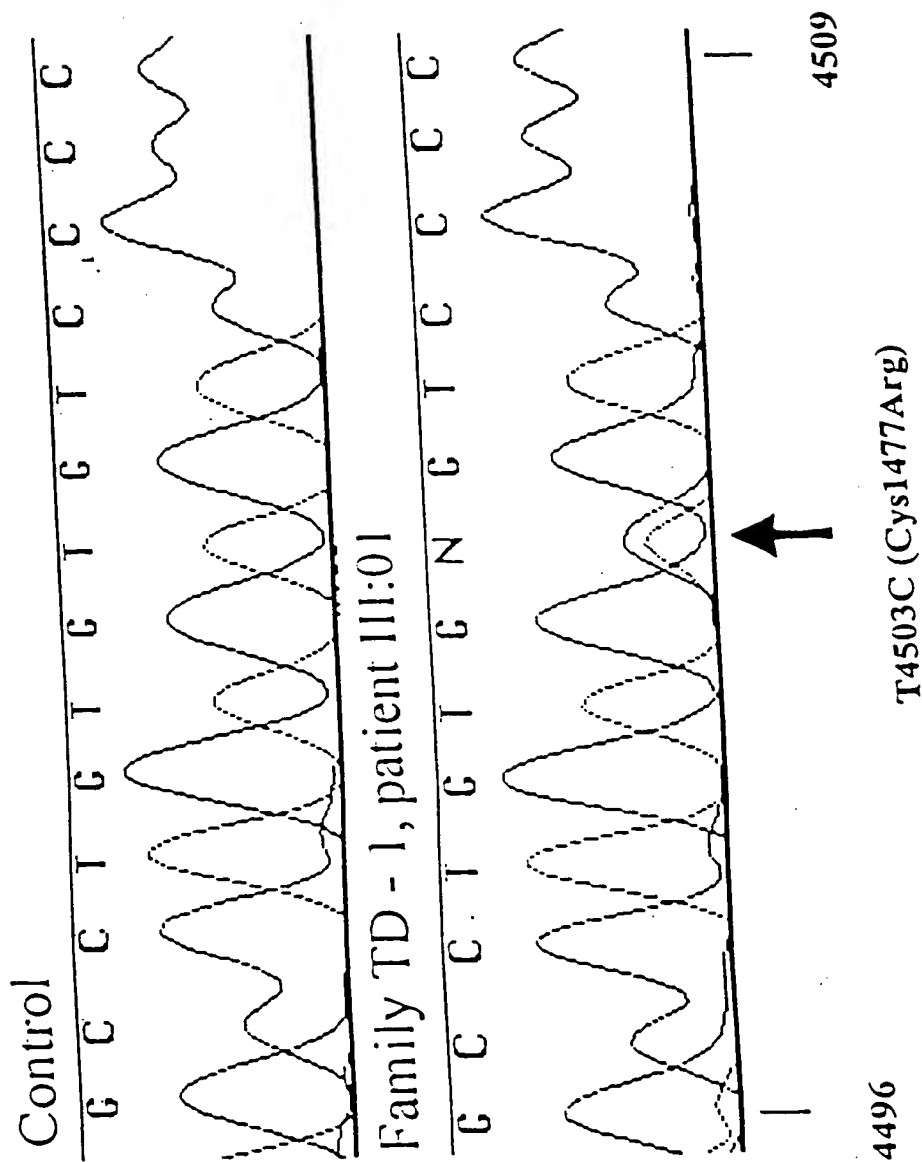


Fig. 4A

Exon 30  
TD-1

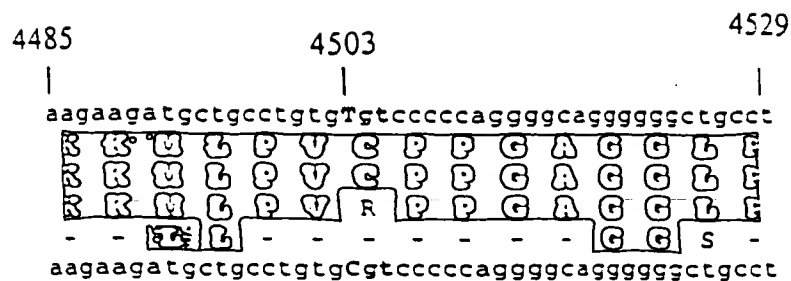
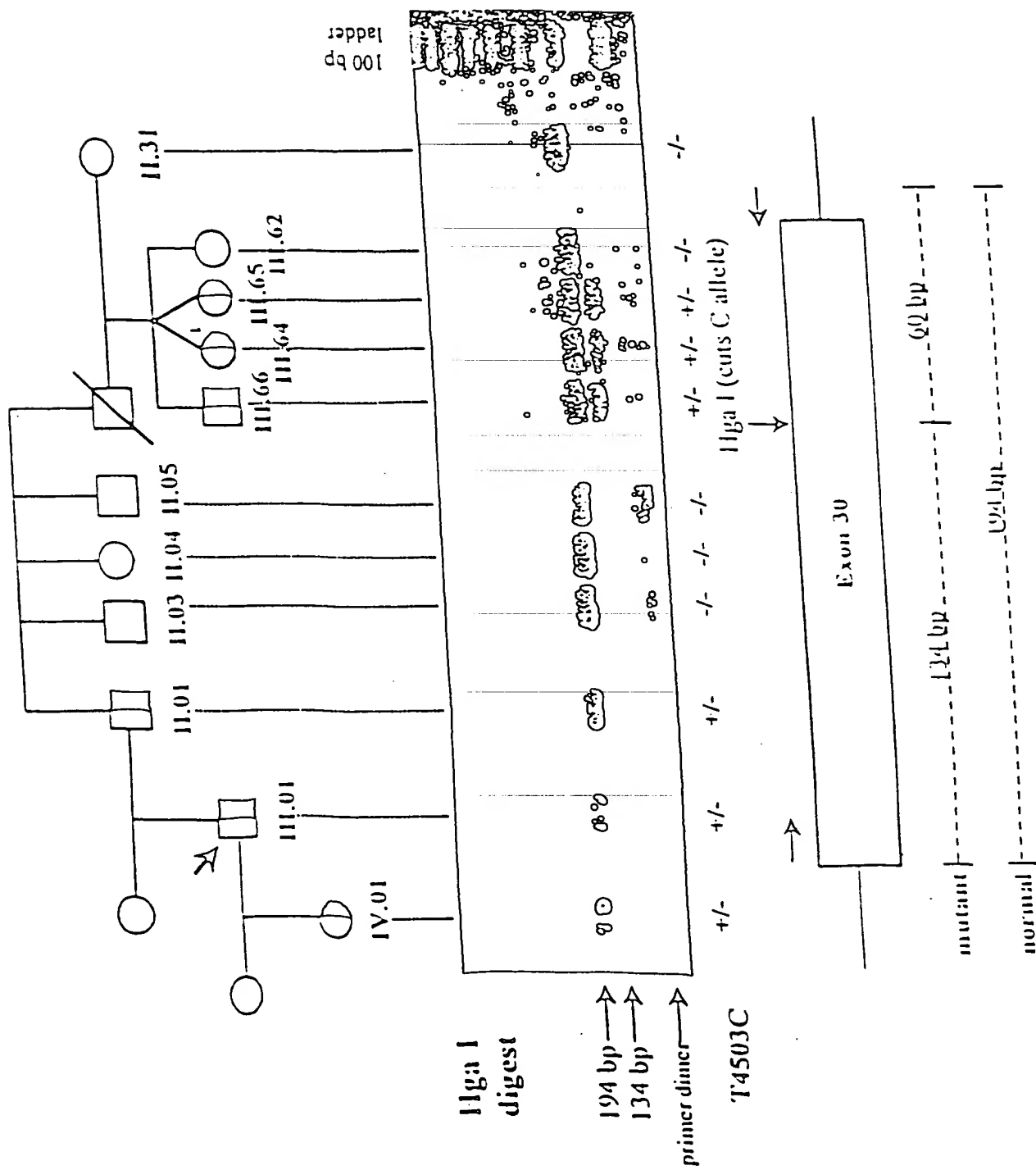


Fig. 4B



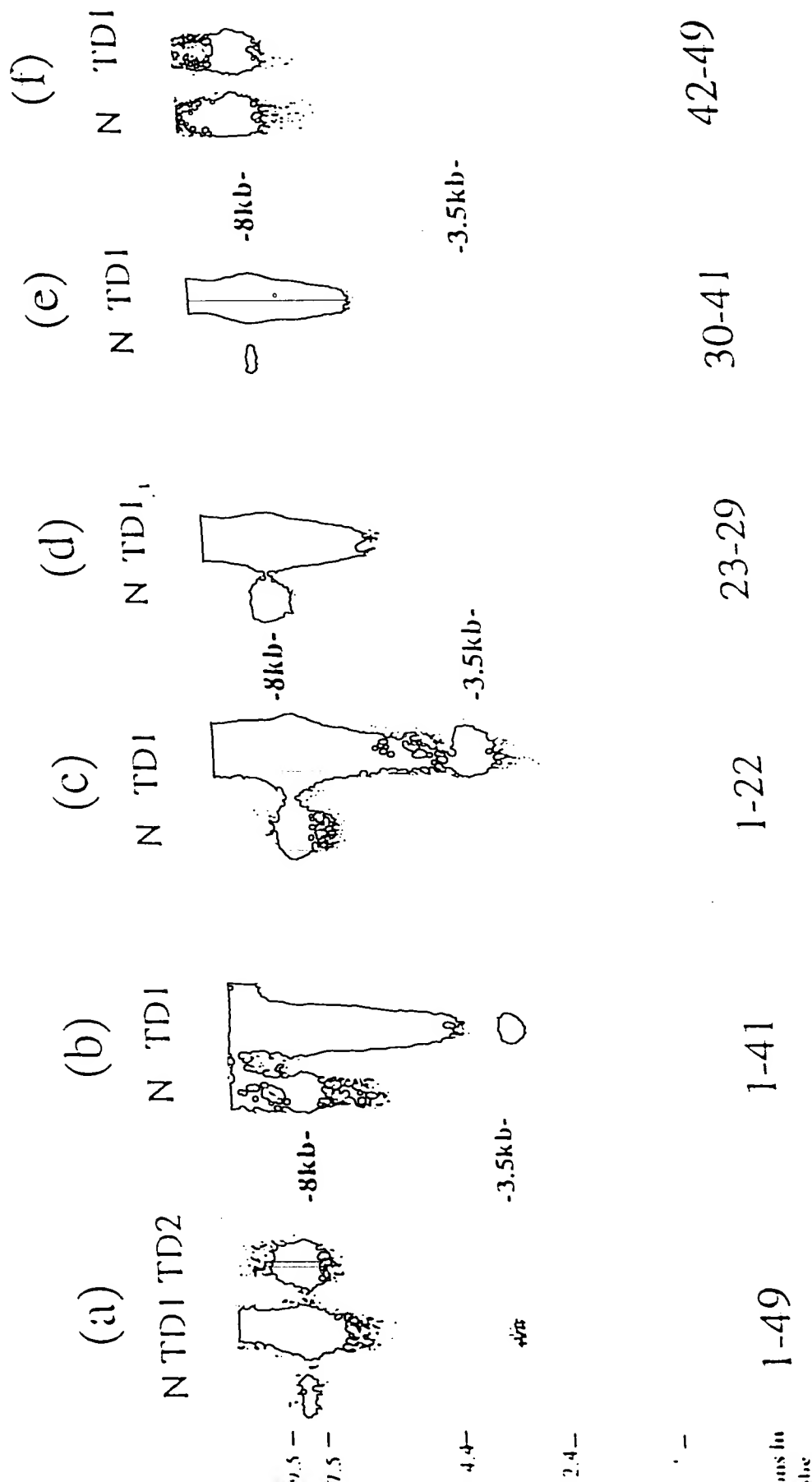


Fig. 4D

# Exon 13 mutation:

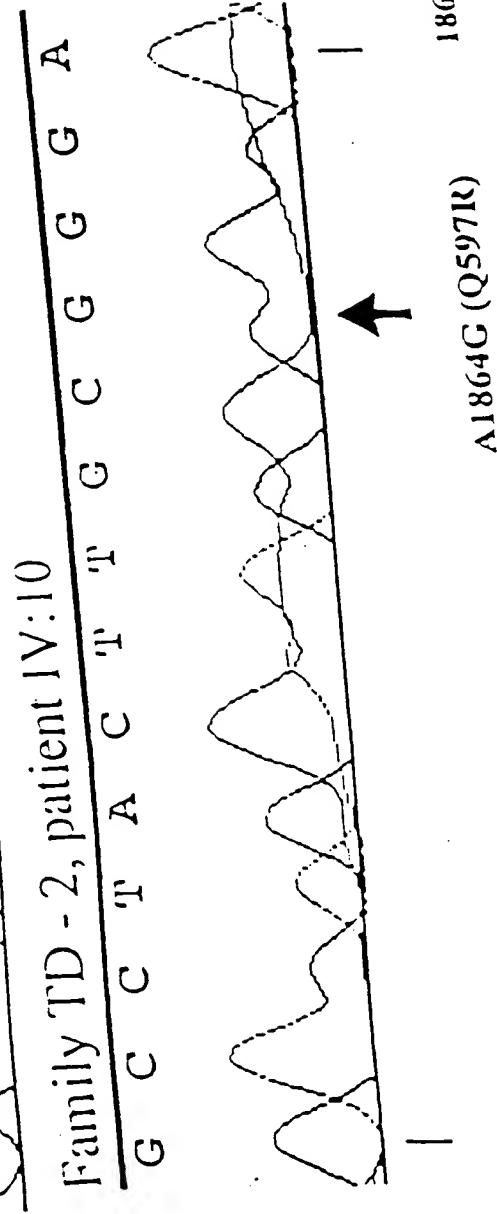
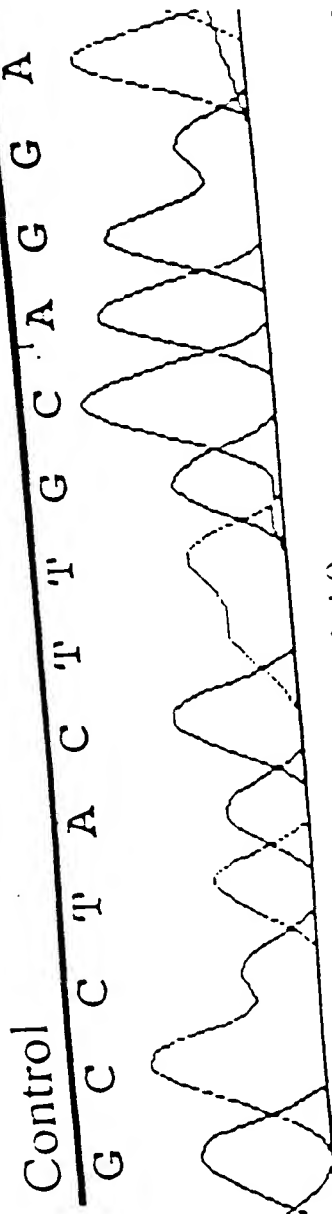


Fig. 5A

005510-01500

Exon 13  
TD-2

1842

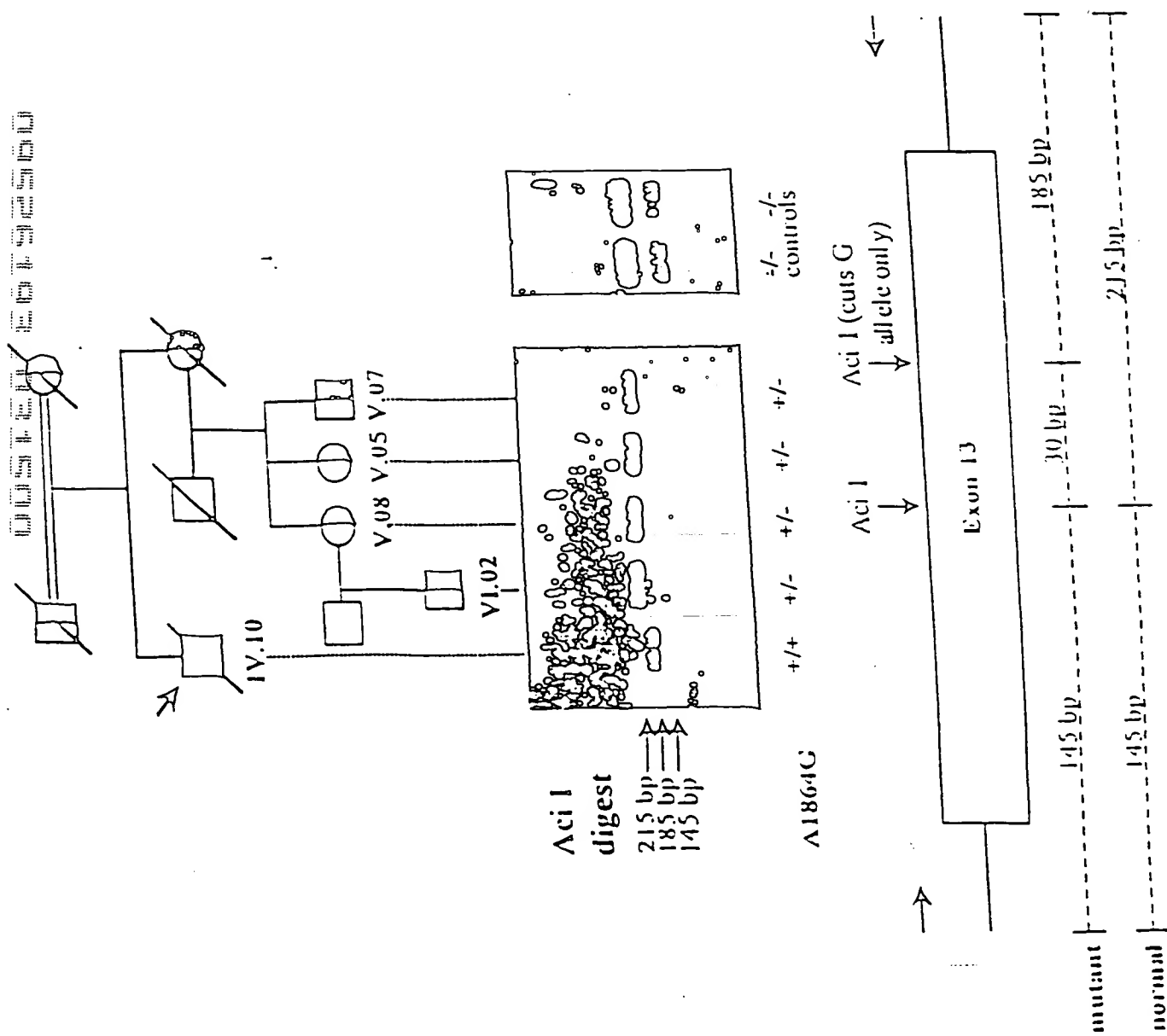
1864

1886

wt sequence  
HUMAN\_ABC1  
MOUSE\_ABC1  
Patient  
CASEL\_ABC  
Patient

```
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N G G F A Y L Q D V V E Q A I
N G G F A Y L Q D V V E Q A I
N G G F A Y L R D V V E Q A I
- - - F M T V Q R A V D V A I
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```

Fig. 5B



# Exon 14: FHA - 1, patient III:01

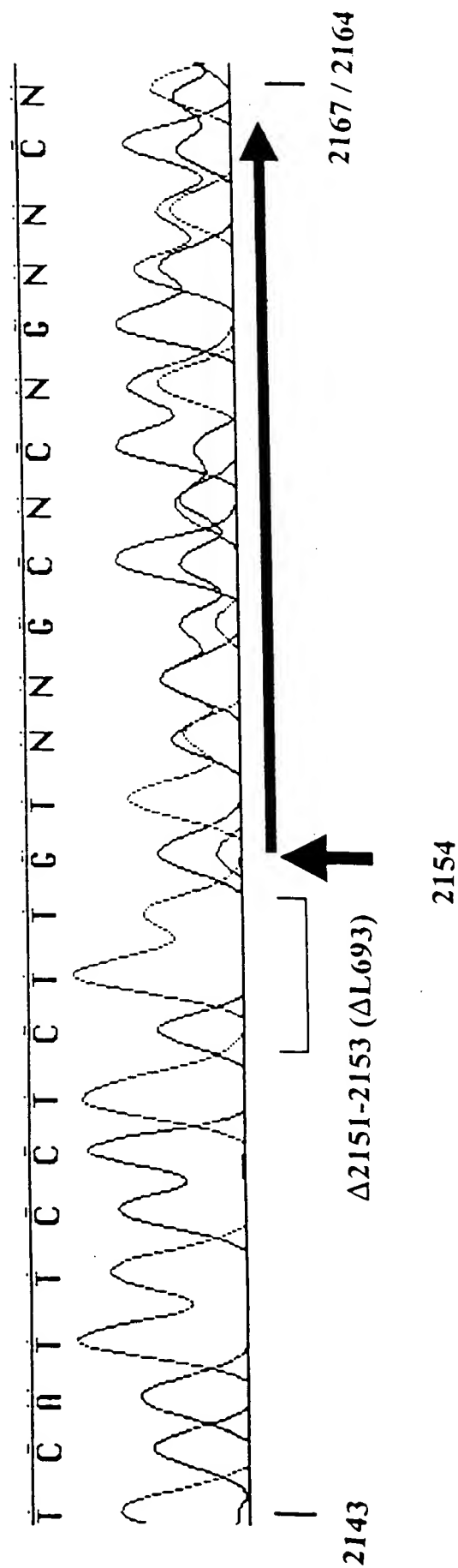


Fig. 6A



Exon 14  
FHA-1

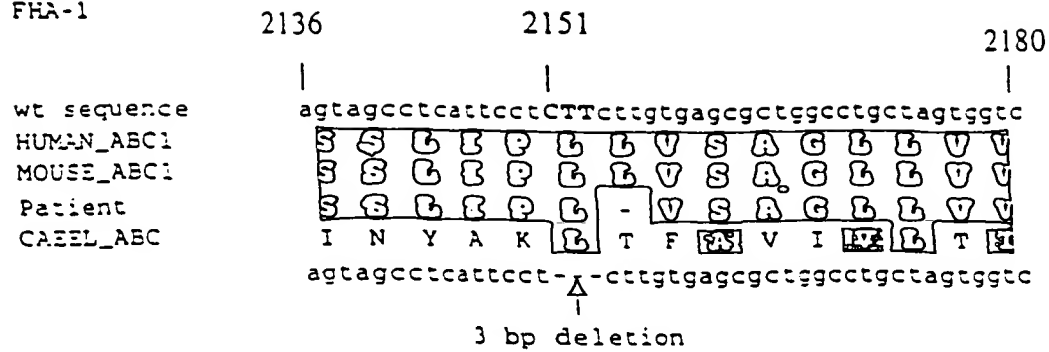


Fig. 6B

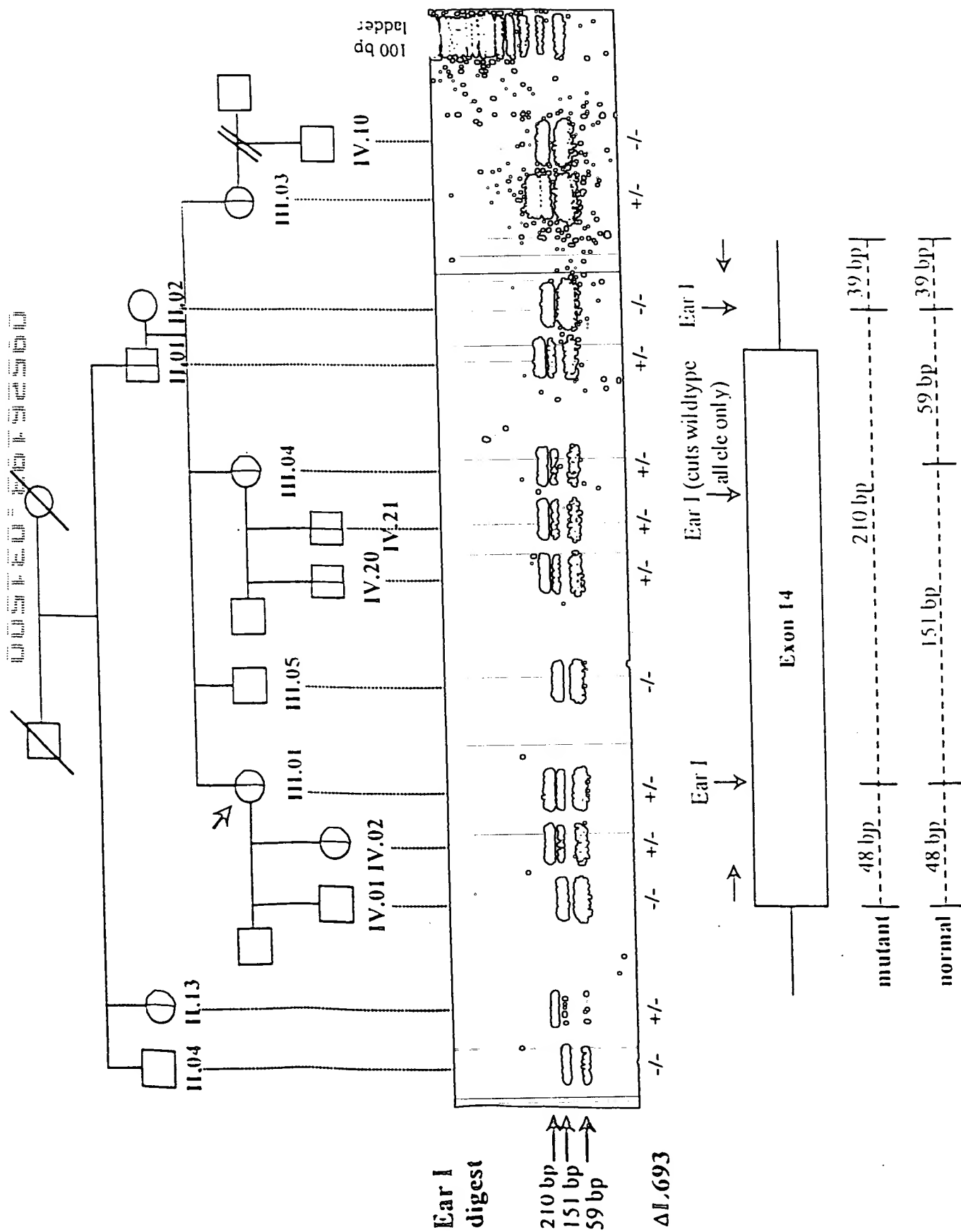


Fig. 6C



Exon 41  
FHA-3

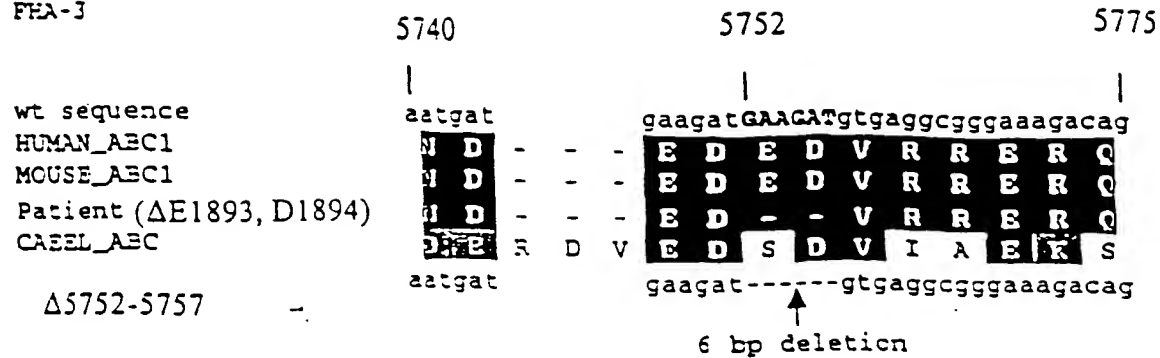
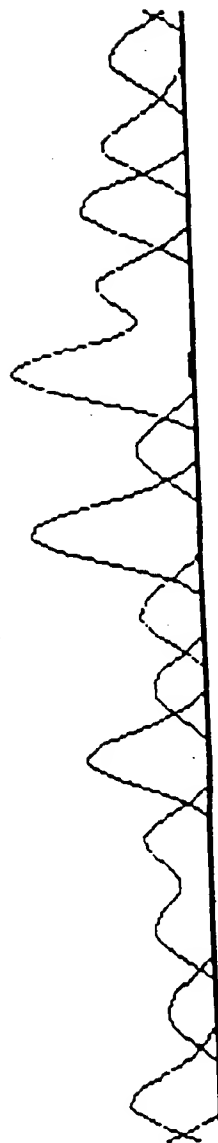


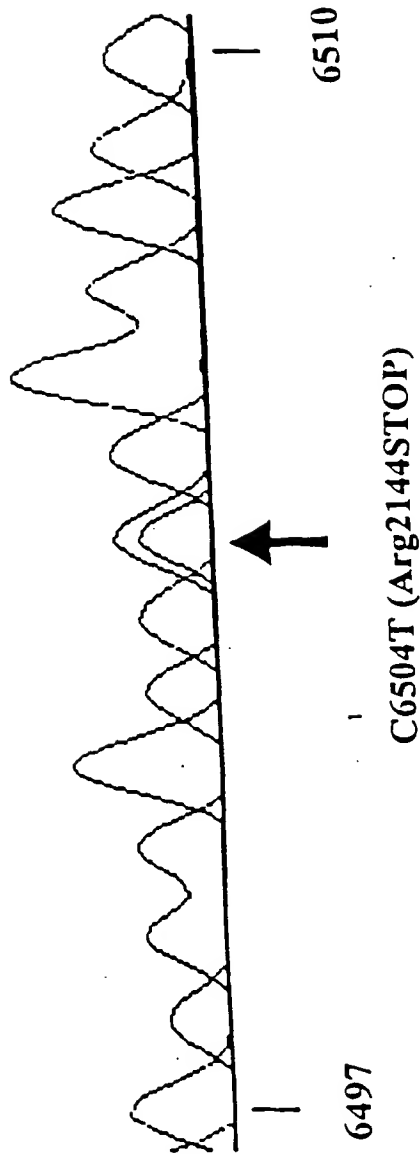
Fig. 6E

Exon 48 mutation:

А С Т Т С Т А С С А И И И И



Family FHA - 2, patient III:01



**Fig. 6f**

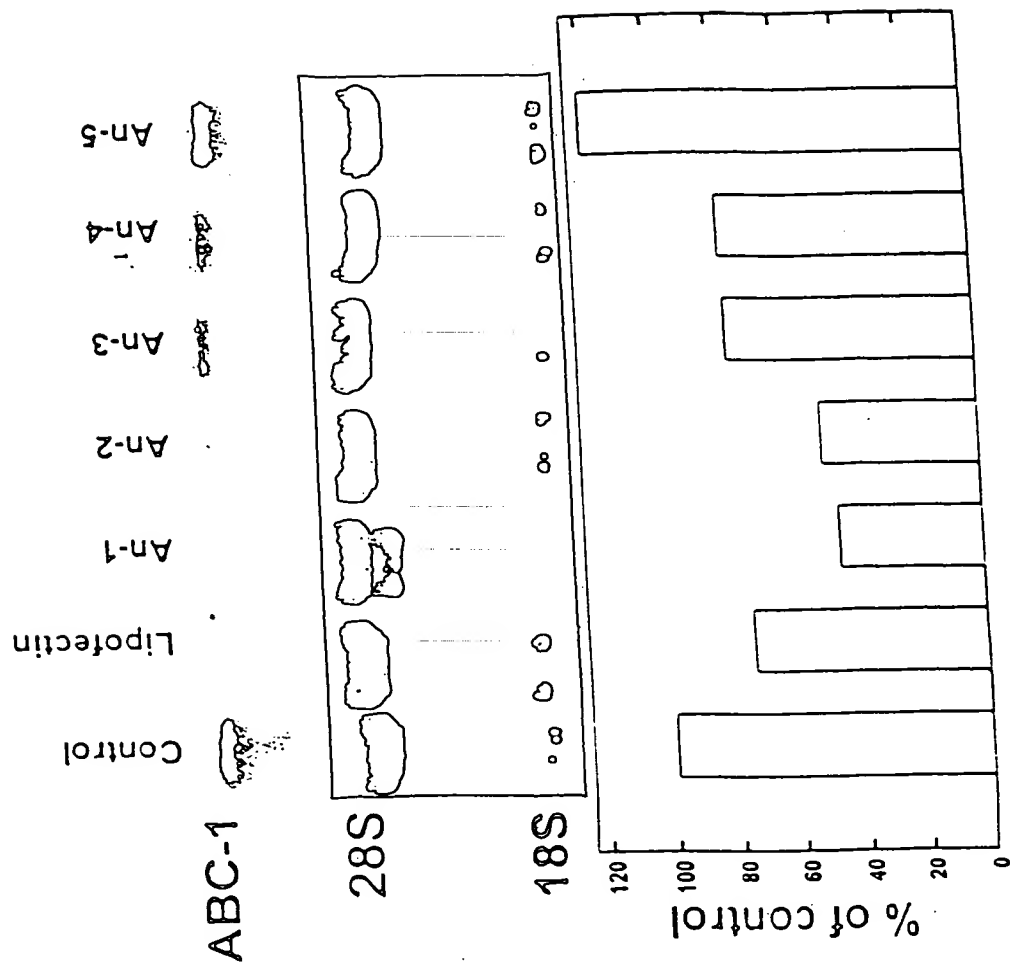


Fig. 7A

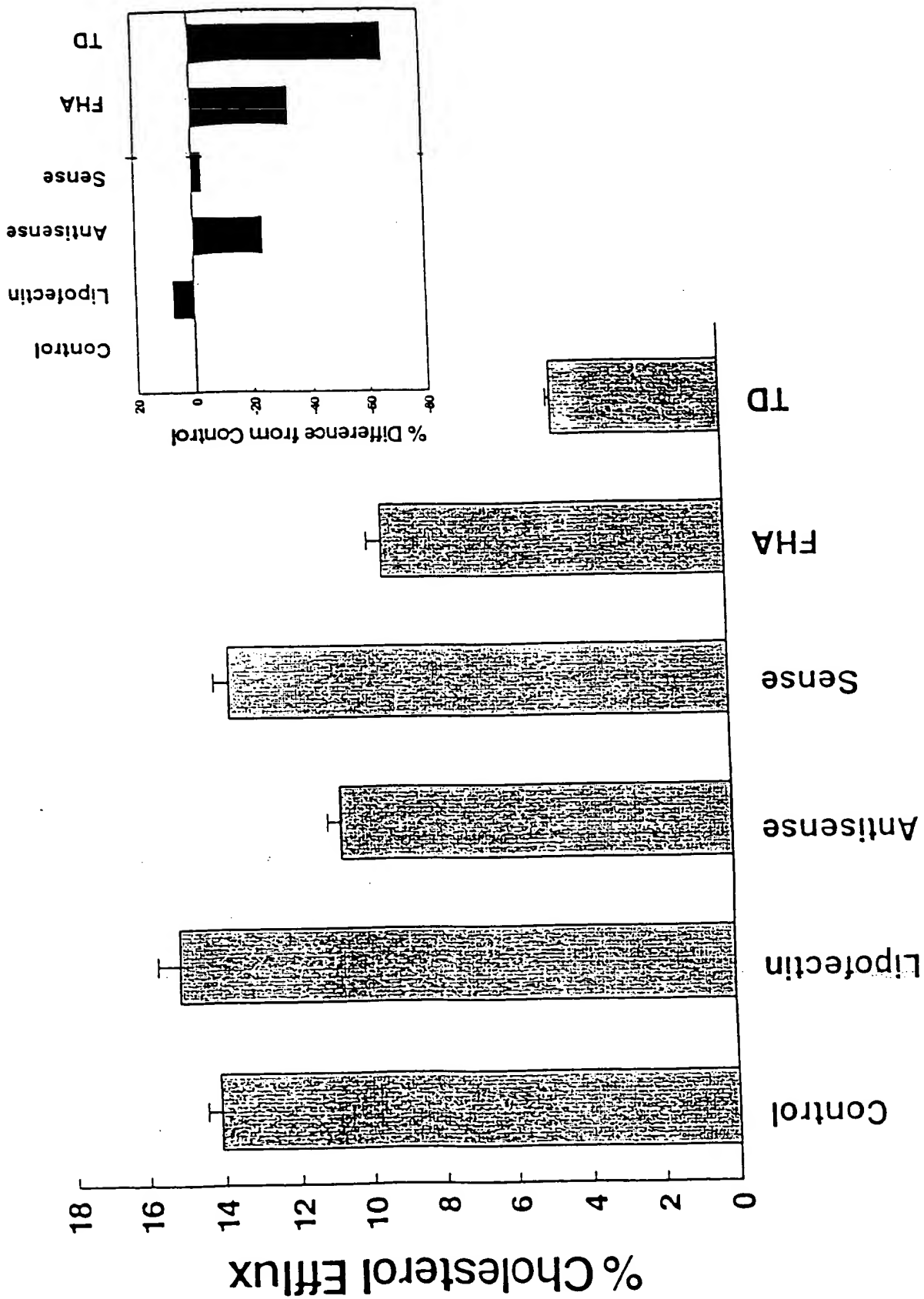


Fig. 7B

Downloaded from www.jci.org

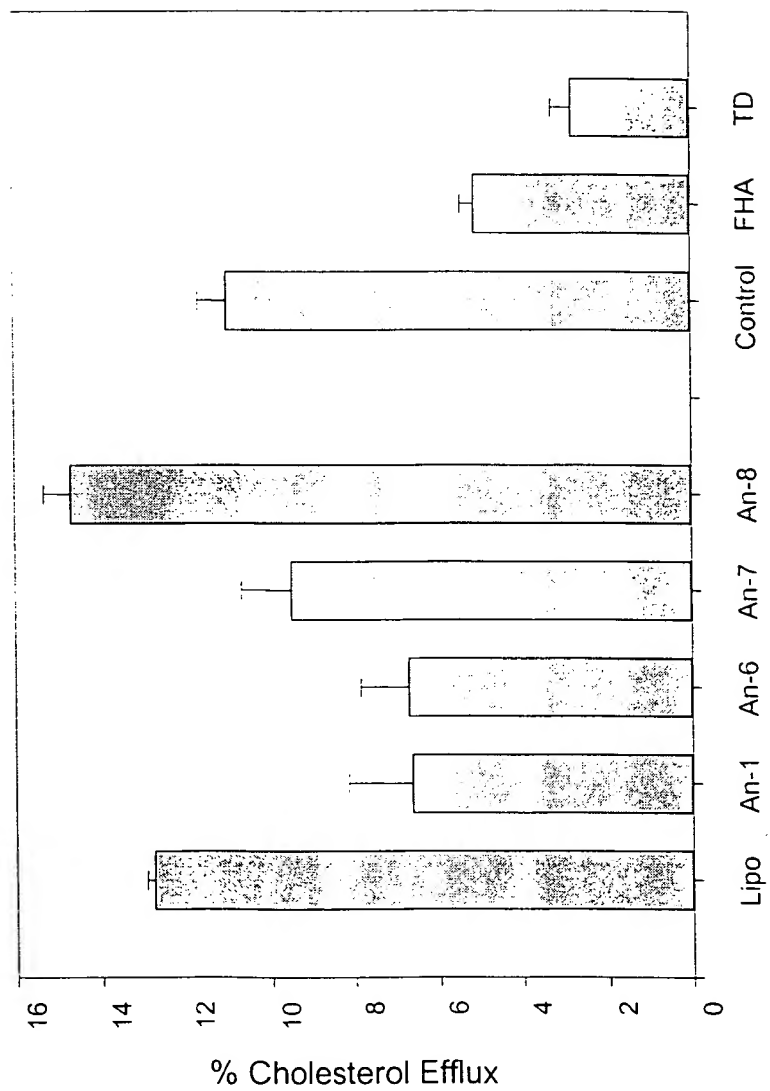


Fig. 7C



## SEQ ID NO: 1

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KLQDFLVDNETFSGFLYHNLSLPKSTVDKMLRADVILHKVFLQGYQLHLTSLCNGSKSEEMIQLGDQEVSE  
LCGLPREKLAAAERVLRNMDILKPILRTLNSTSPFPSKELAEATKTLHSLGTLAQELFSMRWSWDMRQE  
VMFLTNNVSSSSSTQIYQAVSRIVCGHPEGGLKIKSLNWYEDNNYKALFGGNGTEEDAETFYDNSTTPYC  
NDLMKNLESSPLSRIIWKALKPLLVGKILYTPDTPATRQVMAEVNKTQELAVFHDLEGMWHEELSPKIWTF  
MENSQEMDLVRMLLDSRDNDHFWEQQLDGLDWTAQDIVAFLAKHPEDVQSSNGSVYTWREAFNETNQAIRT  
ISRFMECVNLNKLEPIATEVWLINKSMELLDERKFWAGIVFTGITPGSIELPHHVKYKIRMDIDNVERTNK  
IKDGYWDPGPRADPFEDMRYVWGGFAYLQDVVEQAIIRVLTGTEKKTGVYMQMPYPICYVDDIFLRVMSRS  
MPLFMTLAWIYSVAVIIKGIVYEKEARLKETMRIMGLDNSILWFSWFISSLIPLLVSAGLLVVILKLGILL  
PYSDPSVVFVFLSVFAVVTILQCFLISTLFSRANLAAACGGIIYFTLYLPVLCVAWQDYVGFTLKIFASL  
LSPVAFGFGCEYFALFEEQGIGVQWDNLFESPVEEDGFNLTTSVSMMLFDTFLYGVMTWYIEAVFPGQYGI  
PRPWYFPCTKSYWFGESDEKSHPGSNQKRRISEICMEEEPHLKLGVSIGNLVKVYRDGMKVAVDGLALNF  
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WFIARLKLSEKHVKAEMEOMALDVLGPSSKLKSKTSQLSGGMQRKLSVALAFVGGSKVVILDEPTAGVDP  
YSRRGIWELLLKYRQGRTIILSTHMHDEADVLDGRIAIISHGKLCCVGSSSLFLKNQLGTGYLTLVKKDVE  
SSLSSCRNSSSTVSYLKKEDSVSQSSSDAGLGSDESHTLTIDVSAISNLIRKHVSEARLVEDIGHELTYV  
LPYEAKEGAFVELFHEIDRLSDLGISSYGISETTLEEIFLKVAEESGVDAETSDGTLPARRNRRAFGDK  
QSCLRPFTEDDAADPNDSIDPESRETDLLSGMDGKGSYQVKGWKLQQQFVALLWKRLLIARRSRKGFFA  
QIVLPAVFVCIALVFSILVPPFGKYPSELELQPMYNEQYTFVSNDAPEDTGTLELLNALTDPGFGTRCME  
GNPIPDTPCQAGEEEWTTAPVPQTIMDLFQNGNWTMNPSPACQCSSDKIKKMLPVCPPGAGGLPPPQRKQ  
NTADILQDLTGRNISDYLVKTYVQIIAKSLKNKIWNVEFRYGGFSLGVSNTQALPPSQEVNDAIKQMKKHL  
KLAkdSSADRFLNSLGRFMTGLDTRNNVKVWFNNKGWHAISSFLNVINNAILRANLQKGENPSHYGITAFN  
HPLNLTKQLSEVALMTTSVDVLVSICVIFAMSFVPASFVFLIQERVSKAKHLQFISGVKPVIIYWLSNFV  
WDMCNVVPATLVIIIFICFQOKSYVSSSTNLPVLALLLLLYGWSITPLMYPASFVFKIPSTAYVVLTSVNL  
FIGINGSVATFVLELFTDNKLNINDILKSVFLIFPHFCLGRGLIDMVKNQAMADALERFGENRFSPLSW  
DLVGRNLFAMAVEGVVFFLITVLIQYRFFIRPRPVNAKLSPLNDEDEDVRRERQRILDGGGQNDILEIKEL  
TKIYRRKRKPAVDRIKVGIPPGECFGLLVNGAGKSSTFKMLTGDTTVTRGDAFLNKNLSILSNIHEVHQNM  
GYCPQFDAITELLTGREHVEFFALLRGVPEKEVGKVGWAIKRLGLVKYGEKYAGNYSGGNKRKLSTAMAL  
IGGPPVVFLDEPTTGMDPKARRFLWNCALSVVKEGRSVVLTSHSMEECEALCTRMAIMVNGRFRCLGVSQH  
LKNRFGDGYTIVVRIAGSNPDLKPVQDFFGLAFPGSVLKEKHRNMLQYQLPSSLSSLARIFSILSQSKKRL  
HIEDYSVSQTTLDQVFNFAKDQSDDDHLKDLSLHKNQTVVDVAVLTSFLQDEKVKESYV\*

Fig. 9A

SEQ ID NO: 2

GTCCCTGCTGTGAGCTCTGGCCGCTGCCTTCCAGGGCTCCCGAGCCACACGCTGGGGGTG  
CTGGCTGAGGGAACATGGCTTGTGGCCTCAGCTGAGGTTGCTGCTGTGGAAGAACCTCA  
CTTTCAGAAGAAGACAAACATGTCAGCTGTTACTGGAAGTGGCCTGGCCTCTATTTATCT  
TCCTGATCCTGATCTCTGTTTCGGCTGAGCTACCCACCCTATGAACAACATGAATGCCATT  
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ATGCCAACAACCCCTGTTTCCGTTACCCGACTCCTGGGGAGGCTCCCGGAGTTGTTGGAA  
ACTTTAACAATCCATTGTGGCTCGCCTGTTCTCAGATGCTCGGAGGCTTCTTTTATACA  
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GGTTCCTGTATCACAACCTCTCTCTCCCAAAGTCTACTGTGGACAAGATGCTGAGGGCTG  
ATGTCATTCTCCACAAGGTATTTTGAAGGCTACCAGTTACATTGACAAGTCTGTGCA  
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TGAAGCCAATCCTGAGAACACTAACTCTACATCTCCCTTCCCGAGCAAGGAGCTGGCTG  
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GAAGCTGGAGTGACATGCGACAGGAGGTGATGTTTCTGACCAATGTGAACAGCTCCAGCT  
CCTCCACCCAAATCTACCAGGCTGTGTCTCGTATTGTCTGCGGGCATCCCGAGGGAGGGG  
GGCTGAAGATCAAGTCTCTCAACTGGTATGAGGACAACAACCTACAAAGCCCTCTTTGGAG  
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AGCCGCTGCTCGTTGGGAAGATCCTGTATACACCTGACACTCCAGCCACAAGGCAGGTCA  
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TTGTGTTCACTGGAATTACTCCAGGCAGCATTGAGCTGCCCCATCATGTCAAGTACAAGA  
TCCGAATGGACATTGACAATGTGGAGAGGACAAATAAAATCAAGGATGGGTACTGGGACC  
CTGGTCTCGAGCTGACCCCTTTGAGGACATGCGGTACGTCTGGGGGGGCTTCGCCTACT  
TGCAGGATGTGGTGGAGCAGGCAATCATCAGGGTGCTGACGGGCACCGAGAAGAAAAC TG

Fig. 9B

GTGTCTATATGCAACAGATGCCCTATCCCTGTTACGTTGATGACATCTTTCTGCGGGTGA  
TGAGCCGGTCAATGCCCCCTCTTCATGACGCTGGCCTGGATTTACTCAGTGGCTGTGATCA  
TCAAGGGCATCGTGTATGAGAAGGAGGCACGGCTGAAAGAGACCATGCGGATCATGGGCC  
TGGACAACAGCATCCTCTGGTTTAGCTGGTTCATTAGTAGCCTCATTCTCTTCTTGTGA  
GCGCTGGCCTGCTAGTGGTCATCCTGAAGTTAGGAAACCTGCTGCCCTACAGTGATCCCA  
GCGTGCTGTTTGTCTTCTGTCCGTGTTTGCTGTGGTGACAATCCTGCAGTGCTTCCTGA  
TTAGCAACTCTTCTCCAGAGCCAACCTGGCAGCAGCCTGTGGGGGCATCATCTACTTCA  
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AGATCTTCGCTAGCCTGCTGTCTCCTGTGGCTTTTGGGTTTGGCTGTGAGTACTTTGCCC  
TTTTTGAGGAGCAGGGCATTGGAGTGCAGTGGGACAACCTGTTTGAGAGTCTGTGGAGG  
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GGTATTTTCTTGCACCAAGTCCTACTGGTTTGGCGAGGAAAGTGATGAGAAGAGCCACC  
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AGCTGGGCGTGTCCATTGAGAACCTGGTAAAGTCTACCGAGATGGGATGAAGGTGGCTG  
TCGATGGCCTGGCACTGAATTTTATGAGGGCCAGATCACCTCCTTCTGGGCCACAATG  
GAGCGGGGAAGACGACCACCATGTCAATCCTGACCGGGTGTTCCTCCCGACCTCGGGCA  
CCGCCTACATCCTGGGAAAAGACATTCGCTCTGAGATGAGCACCATCCGGCAGAACCTGG  
GGGTCTGTCCCCAGCATAACGTGCTGTTTGACATGCTGACTGTGGAAGAACACATCTGGT  
TCTATGCCCCGCTTGAAAGGGCTCTCTGAGAAGCACGTGAAGGCGGAGATGGAGCAGATGG  
CCCTGGATGTTGGTTTGCCATCAAGCAAGCTGAAAAGCAAAACAAGCCAGCTGTCAGGTG  
GAATGCAGAGAAAGCTATCTGTGGCCTTGGCCTTTGTGCGGGGATCTAAGGTTGTCAATC  
TGGATGAACCCACAGCTGGTGTGGACCCTTACTCCCGCAGGGGAATATGGGAGCTGCTGC  
TGAAATACCGACAAGGCCGCACCATTATTCTCTCTACACACCACATGGATGAAGCGGACG  
TCCTGGGGGACAGGATTGCCATCATCTCCCATGGGAAGCTGTGCTGTGTGGGCTCCTCCC  
TGTTTCTGAAGAACCAGCTGGGAACAGGCTACTACCTGACCTTGGTCAAGAAAGATGTGG  
AATCCTCCCTCAGTTCCTGCAGAAACAGTAGTAGCACTGTGTCAATACCTGAAAAAGGAGG  
ACAGTGTTTCTCAGAGCAGTTCCTGATGCTGGCCTGGGCAGCGACCATGAGAGTGACACGC  
TGACCATCGATGTCTCTGCTATCTCCAACCTCATCAGGAAGCATGTGTCTGAAGCCCGGC  
TGGTGGAAGACATAGGGCATGAGCTGACCTATGTGCTGCCATATGAAGCTGCTAAGGAGG  
GAGCCTTTGTGGAACCTCTTTCATGAGATTGATGACCGCTCTCAGACCTGGGCATTTCTA  
GTTATGGCATCTCAGAGACGACCCTGGAAGAAATATTCCTCAAGGTGGCCGAAGAGAGTG  
GGGTGGATGCTGAGACCTCAGATGGTACCTTGCCAGCAAGACGAAACAGGCGGGCCTTCG  
GGGACAAGCAGAGCTGTCTTCGCCCCTTCACTGAAGATGATGCTGCTGATCCAAATGATT

Fig. 9C

CTGACATAGACCCAGAATCCAGAGAGACAGACTTGCTCAGTGGGATGGATGGCAAAGGGT  
CCTACCAGGTGAAAGGCTGGAACTTACACAGCAACAGTTTGTGGCCCTTTTGTGGAAGA  
GACTGCTAATTGCCAGACGGAGTCGGAAAGGATTTTTTGTCTCAGATTGTCTTGCCAGCTG  
TGTTTGTCTGCATTGCCCTTGTGTTTCAGCCTGATCGTGCCACCCTTTGGCAAGTACCCCA  
GCCTGGAACCTTCAGCCCTGGATGTACAACGAACAGTACACATTTGTCTCAGCAATGATGCTC  
CTGAGGACACGGGAACCTTGGAACCTTTAAACGCCCTCACCAAAGACCCTGGCTTCGGGA  
CCCGCTGTATGGAAGGAAACCCAATCCCAGACACGCCCTGCCAGGCAGGGGAGGAAGAGT  
GGACCACTGCCCCAGTTCCCCAGACCATCATGGACCTCTTCCAGAATGGGAACTGGACAA  
TGCAGAAACCTTCACCTGCATGCCAGTGTAGCAGCGACAAAATCAAGAAGATGCTGCCTG  
TGTGTCCCCCAGGGGCAGGGGGGCTGCCTCCTCCACAAAGAAAACAAAACACTGCAGATA  
TCCTTCAGGACCTGACAGGAAGAAACATTTTCGGATTATCTGGTGAAGACGTATGTGCAGA  
TCATAGCCAAAAGCTTAAAGAACAAGATCTGGGTGAATGAGTTTAGGTATGGCGGCTTTT  
CCCTGGGTGTCTAGTAATACTCAAGCACTTCCTCCGAGTCAAGAAGTTAATGATGCCATCA  
AACAAATGAAGAAACACCTAAAGCTGGCCAAGGACAGTTCTGCAGATCGATTTCTCAACA  
GCTTGGGAAGATTTATGACAGGACTGGACACCAGAAATAATGTCAAGGTGTGGTTCAATA  
ACAAGGGCTGGCATGCAATCAGCTCTTTCCTGAATGTCATCAACAATGCCATTCTCCGGG  
CCAACCTGCAAAAAGGGAGAGAACCCTAGCCATTATGGAATTACTGCTTTCAATCATCCCC  
TGAATCTCACCAAGCAGCAGCTCTCAGAGGTGGCTCTGATGACCACATCAGTGGATGTCC  
TTGTGTCCATCTGTGTCTCTTTGCAATGTCCTTCGTCCCAGCCAGCTTTGTCTGATTCC  
TGATCCAGGAGCGGGTCAGCAAAGCAAAACACCTGCAGTTCATCAGTGGAGTGAAGCCTG  
TCATCTACTGGCTCTCTAATTTTGTCTGGGATATGTGCAATTACGTTGTCCCTGCCACAC  
TGGTCATTATCATCTTCATCTGCTTCCAGCAGAAGTCCTATGTGTCTCCACCAATCTGC  
CTGTGCTAGCCCTTCTACTTTTGTCTGTATGGGTGGTCAATCACACCTCTCATGTACCCAG  
CCTCCTTTGTGTTCAAGATCCCCAGCACAGCCTATGTGGTGCTCACCAGCGTGAACCTCT  
TCATTGGCATTAAATGGCAGCGTGGCCACCTTTGTGCTGGAGCTGTTACCGACAATAAGC  
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GACGAGGGCTCATCGACATGGTGAAAAACCAGGCAATGGCTGATGCCCTGGAAAGGTTTG  
GGGAGAATCGCTTTGTGTACACATTATCTTGGGACTTGGTGGGACGAAACCTCTTCGCCA  
TGGCCGTGGAAGGGGTGGTGTCTTCTCCTCATTACTGTTCTGATCCAGTACAGATTCTTCA  
TCAGGCCCAGACCTGTAAATGCAAAGCTATCTCCTCTGAATGATGAAGATGAAGATGTGA  
GGCGGGAAAGACAGAGAATTCTTGATGGTGGAGGCCAGAATGACATCTTAGAAATCAAGG  
AGTTGACGAAGATATATAGAAGGAAGCGGAAGCCTGCTGTTGACAGGATTTGCGTGGGCA  
TTCCTCCTGGTGAGTGCTTTGGGCTCCTGGGAGTTAATGGGGCTGGAAAATCATCAACTT  
TCAAGATGTTAACAGGAGATACCACTGTTACCAGAGGAGATGCTTTCCTTAACAAAATA

Fig. 9D

GTATCTTATCAAACATCCATGAAGTACATCAGAACATGGGCTACTGCCCTCAGTTTGATG  
CCATCACAGAGCTGTTGACTGGGAGAGAACACGTGGAGTTCTTTGCCCTTTTGAGAGGAG  
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AGTATG3AGAAAAATATGCTGGTAACTATAGTGGAGGCAACAAACGCAAGCTCTCTACAG  
CCATGGCTTTGATCGGCGGGCCTCCTGTGGTGTTCCTGGATGAACCCACCACAGGCATGG  
ATCCCAAGCCCCGGCGGTTCTTGTGGAATTGTGCCCTAAGTGTGTCAAGGAGGGGAGAT  
CAGTAGTGCTTACATCTCATAGTATGGAAGAATGTGAAGCTCTTTGCACTAGGATGGCAA  
TCATGCTCAATGGAAGGTTTCAGGTGCCTTGGCAGTGTCCAGCATCTAAAAAATAGGTTTG  
GAGATGTTTATACAATAGTTGTACGAATAGCAGGGTCCAACCCGGACCTGAAGCCTGTCC  
AGGATTTCTTTGGACTTGCATTTCTTGGAAAGTGTCTAAAAGAGAAACACCGGAACATGC  
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TATTTGTGAACTTTGCCAAGGACCAAGTGATGATGACCACTTAAAAGACCTCTCATTAC  
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TGAAAGAAAGCTATGTATGAAGAATCCTGTTTCATACGGGGTGGCTGAAAGTAAAGAGGAA  
CTAGACTTTCCTTTGCACCATGTGAAGTGTGTGGAGAAAAGAGCCAGAAGTTGATGTGG  
GAAGAAGTAAACTGGATACTGTACTGATACTATTCAATGCAATGCAATTCAATGCAATGA  
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CACATCCATTGCTGGCAATGAGTGTGCCAGAGTTATTAGTGCCAAGTTTTTCAGAAAGTT  
TGAAGCACCATGGTGTGTCTGCTCACTTTTGTGAAAGCTGCTCTGCTCAGAGTCTATCA  
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CTGCTGGGGCTGCAACTGCTGAAGCCAGGGCATGGGATTAAAGAGATTGTGCGTTCAAAC  
CTAGGGAAGCCTGTGCCCATTTGTCTGACTGTCTGCTAACATGGTACACTGCATCTCAA  
GATGTTTATCTGACACAAGTGTATTATTTCTGGCTTTTTGAATTAATCTAGAAAATGAAA

Fig. 9E

Exon Forward Primer (bp)		SEQ ID No.		Reverse Primer		SEQ ID No.		Intron(kb)		Intron (kb)	
exon 1	140	GGCTGGATAGCAGTCC1CA	70	A1CCCCAATCAAAACACCA	119	intron 1	>6.413	intron 1	>6.413	intron 1	>6.4
exon 2	94	GGATTTCCAGATCCCGT	71	AAGTCCCAATTTAGCCAGGTT	120	intron 2	>4.241	intron 2	>4.241	intron 2	>4.2
exon 3	142	GACAGACTGGCATGAAGCA	72	CAGCCACTTGAATTTCTCC	121	intron 3	>1.248 (1.6)	intron 3	>1.248 (1.6)	intron 3	1.6
exon 4	119	GCATTTGGCAGTCACTTCTG	73	GGTGCAGGTCATTTTCCAAT	122	intron 4	>1.512	intron 4	>1.512	intron 4	>1.5
exon 5	122	CGTTTCCACCTGCTCCCT	74	CCCTTCCACCACTTACAA	123	intron 5	>1.769 (3)	intron 5	>1.769 (3)	intron 5	3
exon 6	177	ACTCAAGACCCAGCTCC	75	TGTTCAAGGAAAGCCCTAC	124	intron 6	>2.726 (10)	intron 6	>2.726 (10)	intron 6	10
exon 7	93	TCGGTTCTTGTTTAACTCA	76	AGSACCTCTTGCAGACTCA	125	intron 7	4.957	intron 7	4.957	intron 7	5
exon 8	241	TCCCAAGGCTTTGAGATGAC	77	AGGAGATGACAGGCGCAAG	126	intron 8	>2.311 (2.5)	intron 8	>2.311 (2.5)	intron 8	2.5
exon 9	140	GGCTCAAAAGCCCTTGTA	78	AGGCACACCTCTGAAGCTACC	127	intron 9	0.332	intron 9	0.332	intron 9	0.3
exon 10	117	GCTGCTGTGATGGGTATCT	79	ACCTCACTACACCTGGGAA	128	intron 10	4.208	intron 10	4.208	intron 10	4.2
exon 11	198	TTTGTAATTTGTAGTCTCTCA	80	GCCTCCTGCTGACCTTAT	129	intron 11	0.747	intron 11	0.747	intron 11	0.7
exon 12	206	TAGTCAGCCCTTGCCCTCTA	81	CAAAATCATGACCAAGTTGAG	130	intron 12	0.523	intron 12	0.523	intron 12	0.5
exon 13	177	AAAGGGGCTTGGTAAGGTA	82	CATGCACATGCACACACATA	131	intron 13	1.787	intron 13	1.787	intron 13	1.8
exon 14	223	GATGTGGTCTCCTCTAGC	83	CGTAGCCGCTGTTGAGCTA	132	intron 14	1.747	intron 14	1.747	intron 14	1.7
exon 15	222	CAAGTGAAGTCTTGGGATTG	84	TGCTTTTATCAGGACTCCA	133	intron 15	1.059	intron 15	1.059	intron 15	1.1
exon 16	205	GCAATTCAAAATTTCTCCAGG	85	CCCATGCACTGCAGAGATTCC	134	intron 16	1.105	intron 16	1.105	intron 16	1.1
exon 17	114	TCAAGGAGGAATGGACCTG	86	AAGGCAGGAGACATCGCTT	135	intron 17	1.798	intron 17	1.798	intron 17	1.8
exon 18	172	CTGAAGATTCAAGCGCAGTG	87	GGGATCAGCATGGTTTCTCTA	136	intron 18	0.99	intron 18	0.99	intron 18	1
exon 19	132	TGCAGACTGAATGGGACATC	88	GCTTAAGTCCCACTCTCTCC	137	intron 19	1.307	intron 19	1.307	intron 19	1.3
exon 20	143	GCCAGGGACACTGTATTCT	89	ATTTTCTCCCACTGTGTGT	138	intron 20	0.204	intron 20	0.204	intron 20	0.2
exon 21	138	AGTCTCTCTGCCCTCACTCA	90	TCACAGAAGCTTAGCCATGA	139	intron 21	0.706	intron 21	0.706	intron 21	0.7
exon 22	221	CCAGTGCCTTACCCCTGCTAA	91	AACAGAGCAGGAGATGGTG	140	intron 22	>0.866 (1.7)	intron 22	>0.866 (1.7)	intron 22	1.7
exon 23	73	CACACAACAGACTTCTTGGGA	92	TCTGCACCTCTCTCTCTCTG	141	intron 23	0.986	intron 23	0.986	intron 23	1
exon 24	203	ACCTGGAACAGGTGTGGTGT	93	ACTGGGCCCAACATTAATCA	142	intron 24	1.668	intron 24	1.668	intron 24	1.7
exon 25	49	GGGCTAACATGCCACTCAGTA	94	CTTCCCACTTGCACAAAC	143	intron 25	0.196	intron 25	0.196	intron 25	0.2
exon 26	114	GTTTTTCAGATGGGGAAG	95	GCCTAAAGCCCATCAAAGAA	144	intron 26	1.396	intron 26	1.396	intron 26	1.4
exon 27	149	CACCAAGAAGGAGCATGG	96	TCAGTGCACTCTGGGCATAA	145	intron 27	1.649	intron 27	1.649	intron 27	1.6
exon 28	125	CTGGACTCGTAGGGATTTCG	97	1CTGAAGTCCATTCCTTTGG	146	intron 28	>0.728 (1.4)	intron 28	>0.728 (1.4)	intron 28	1.4
exon 29	99	GCCTGTCACAGAGAAATGCTT	98	CAATGTGGCATGCAAGTTGAT	147	intron 29	>2.589 (3)	intron 29	>2.589 (3)	intron 29	3
exon 30	190	TTACGGAATGATCTGTGCTC	99	GAAGGTACGAGCCCATCTCT	148	intron 30	1.521	intron 30	1.521	intron 30	1.5
exon 31	95	AGTCAGGTTCCGGTCACAC	100	CATTTCGCCCACTGTTTCAG	149	intron 31	>0.944 (1)	intron 31	>0.944 (1)	intron 31	>0.9
exon 32	33	CGTTCCCTTATATCCTCAGGTG	101	CCAAGGCTTTCTCAATCCA	150	intron 32	>1.062 (6.5)	intron 32	>1.062 (6.5)	intron 32	>1.0
exon 33	106	CGTTGACACACTCGCACTGA	102	GATCCGTTTAAACCTGCCAAC	151	intron 33	1.475	intron 33	1.475	intron 33	1.5
exon 34	75	TGTTGCCACAGGTTCCAGA	103	ATGCCCTGCCCACTTTTAC	152	intron 34	0.522	intron 34	0.522	intron 34	0.5
exon 35	170	TGAGGTTTATGGGCATGGTT	104	CTCTGCAGCTGTCCCTTAC	153	intron 35	1.228	intron 35	1.228	intron 35	1.2
exon 36	178	ATGTTTTCTTCTGCTGTC	105	TATCAATCCATGGCCCTGAC	154	intron 36	>1.898 (2)	intron 36	>1.898 (2)	intron 36	2
exon 37	116	ATCTGCCCTTCTTGTCTGA	106	AGAGTCCCTGCCCTCTCTCT	155	intron 37	0.112	intron 37	0.112	intron 37	0.1
exon 38	145	AGGGAGCTGCACAGTGGATA	107	AAGGCAGTCAGCAGTGTCAA	156	intron 38	1.545	intron 38	1.545	intron 38	1.5
exon 39	124	TCACTCCCATATTTCAAGACTTGA	108	GGGGAACATCTCTGTGCTTAG	157	intron 39	1.087	intron 39	1.087	intron 39	1.1
exon 40	130	TGTTTATTGGAAGATCGGTGAA	109	CCATTGGTGAAGTGTTCCTCT	158	intron 40	0.265	intron 40	0.265	intron 40	0.3
exon 41	121	CGTTAGCACTGAATCTTTGTCCTG	110	AGTCAGCAAACTGTGGGTT	159	intron 41	>0.622 (0.9)	intron 41	>0.622 (0.9)	intron 41	0.9
exon 42	63	AGTCCTGCCCTCCACAGTTG	111	ATTGCTCCATCTGGCATAA	160	intron 42	0.909	intron 42	0.909	intron 42	0.9
exon 43	107	GGTAGTACGTTGTAGGGGCA	112	TCATGGATGATTTTATGTGCTTC	161	intron 43	2.355	intron 43	2.355	intron 43	2.4
exon 44	142	CAGGAACATTAGGCGCAGATTG	113	GGGTGTGGAAAGGCCATAAG	162	intron 44	0.372	intron 44	0.372	intron 44	0.4
exon 45	135	CATGTATGTGTAGGACACATGA	114	GCCAATCATACAACAGCCCT	163	intron 45	>1.059 (1.3)	intron 45	>1.059 (1.3)	intron 45	1.3
exon 46	104	CTGTTTCAAGATGCTTCTGCG	115	TGATCGCATATCTACTTGGAAA	164	intron 46	0.483	intron 46	0.483	intron 46	0.5
exon 47	93	CCTAGGAAGCTGGAATGCTG	116	TCCCTTTATTTTAGAGGACCA	165	intron 47	0.659	intron 47	0.659	intron 47	0.7
exon 48	244	GGGTCCCAAGGGTTCAGTAT	117	GATCAGGAATTCAGACACAA	166	intron 48	0.941	intron 48	0.941	intron 48	0.9
exon 49	295	CTTGACCTAAATTTCAACATCTGG	118	TGGGTCCCATATATAGAGTTTCACA	167		>1.075		>1.075		

Fig. 10

Errors in public sequence (differences between all samples and Genbank entry AJ012376.1):					
Exon/Intron	Nucleotide	Amino acid change		Sequence difference/context	SEQ ID NO:
2	T150C	no change	Public sequence:	TGTCAGCTGTACTGGAAGTGG	168
	A152G		Correct sequence:	TGTCAGCTGCTGCTGGAAGTGG	169
7	C839T	no change	Public sequence:	AGGAGCTGGCGGAAGCCACAA	170
			Correct sequence:	AGGAGCTGGCTGAAGCCACAA	171
33	C4738T	T1495I	Public sequence:	AATGATGCCACCAACAAATG	172
			Correct sequence:	AATGATGCCATCAACAAATG	173
35	C5017T	P1588L	Public sequence:	GAGGTGGCTCCGATGACCACA	174
			Correct sequence:	GAGGTGGCTCTGATGACCACA	175
43	G5995A	R1914K	Public sequence:	TTCTTTAACAGAAATAGTATG	176
			Correct sequence:	TTCTTTAACAAAAATAGTATG	177
48	C6577T	P2108L	Public sequence:	GGAAGTGTTCAAAAGAGAAA	178
			Correct sequence:	GGAAGTGTTCAAAAGAGAAA	179
49	G6899A	not applicable	Public sequence:	AGTAAAGAGGGACTAGACTTT	180
			Correct sequence:	AGTAAAGAGGAACAGACTTT	181
Mutations:					SEQ ID NO:
13	A1864G	Q597R	More common:	GCTTACTGTGAGGATGTGGTG	182
			Less common:	GCTTACTGTGGGATGTGGTG	183
14	delta CTT 2151-3	delta L093	More common:	CCTCATTCCTCTCTGTGAGCG	184
			Less common:	CCTCATTCCTCTCTGTGAGCG	185
15	G2385A	V771M	More common:	GCAGGACTACGTGGCTTCAC	186
			Less common:	GCAGGACTACATGGCTTCAC	187
18	C2799T	R909Stop	More common:	AAAAGTCTACCGAGATGGGAT	188
			Less common:	AAAAGTCTACTGAGATGGGAT	189
18	C2860T	T929I	More common:	GGCCAGATCACTCTCTCTCTG	190
			Less common:	GGCCAGATCATCTCTCTCTCTG	191
22	T3346C	M1091T	More common:	ACACACACATGGATGAAGCG	192
			Less common:	ACACACACACGGATGAAGCG	193
Intron 24	(+1) G to C splice donor site	Altered transcript length	More common:	CCTGGAAGAACTAAGTTAAGT	194
			Less common:	CCTGGAAGAACTAAGTTAAGT	195
30	T4503C	C1477R	More common:	GCTGCTGTGTGTCTCCCCAGG	196
			Less common:	GCTGCTGTGTGTCTCCCCAGG	197
35	GG 4956-57 to C	Frameshift at aa1628	More common:	TAGCCATTATGGAACTACTGCT	198
			Less common:	TAGCCATTATCAACTACTGCT	199
41	delta AAGATG 5752	delta(E.D)1893-1894	More common:	GATGAAGATGAAGATGTGAGGCGGA	200
			Less common:	GATGAAGATG/TGAGGCGGA	201
48	C6504T	R2144Stop	More common:	AATAGTTGTACGAATAGCAGG	202
			Less common:	AATAGTTGTATGAATAGCAGG	203
Promoter Variants:					SEQ ID NO:
Location	Position Relative to Xenon cDNA	Position Relative to SEQ ID NO: 14 Containing Exon 1			
1	G57C	8216	More common:	ACACGCTGCGGCTGCTGCTG	204
			Less common:	ACACGCTGAGCGTGTGCTG	205
5'	(-) 4 ins. G	8158	More common:	GACCAAGCAAGCGTCCCTG	206
			Less common:	GACCAAGCAAGCGTCCCTG	207
5'	A (-) 380 G	7780	More common:	CATTTTCTTAAAGAGAGAGT	208
			Less common:	CATTTTCTTAAAGAGAGAGT	209
5'	A (-) 479 C	7681	More common:	GAAAAATTAGTATGTAAGGAAG	210
			Less common:	GAAAAATTAGTATGTAAGGAAG	211
5'	A (-) 738 G	7422	More common:	CCTCGGCTTGCAGGTTTACGGAAT	212
			Less common:	CCTCGGCTTGCAGGTTTACGGAAT	213
5'	A (-) 1045 G	7115	More common:	TATGTGCTTACCAAGGAGCTTGTT	214
			Less common:	TATGTGCTTACCGTGGAGCTTGTT	215
5'	A (-) 1113 G	7047	More common:	GTGACACCAAGGAGTAGGG	216
			Less common:	GTGACACCAAGGAGTAGGG	217
5'	(-) 1181 ins. CCCT	5679	More common:	AGTATCCCTTGTTCACGAGAA	218
			Less common:	AGTATCCCTTGTTCACGAGAA	219

Fig. 11

Polymorphisms: Exon/Intron	Nucleotide#	Amino acid change		Sequence difference/context	SEQ ID NO:
5	G548A	no change	More common: Less common:	CTGGGTTCCGTATCAAACT CTGGGTTCCGTATCAAACT	220 221
5	G730A	R219K	More common: Less common:	GGCCTACCAAGGAGAAACT GGCCTACCAAGGAGAAACT	222 223
Intron 7	G (+) 2383 T	Not applicable	Allele 1: Allele 2:	TTTAAAGGGGGTGAATTAGGA TTTAAAGGGGGTGAATTAGGA	224 225
Intron 7	G (+) 3035 T	Not applicable	Allele 1: Allele 2:	GAAGAAATTTGTTTTTGAAT GAAGAAATTTTGTGTTGAAT	226 227
8	C1010T	no change	More common: Less common:	GCGGGCATCCGAGGGAGGG GCGGGCATCCGAGGGAGGG	228 229
9	G1022A	no change	More common: Less common:	AGGGAGGGGGGCTGAAGATCA AGGGAGGGGGGCTGAAGATCA	230 231
Intron 9	(-) 42 ins. G	Not applicable	More common: Less common:	AGGAGCCAAAGCTCATTTG AGGAGCCAAAGCTCATTTG	232 233
Intron 13	T (+) 24 A	Not applicable	More common: Less common:	AAGCCACTGTTTTTAACCAAT AAGCCACTGTTTTTAACCAAT	234 235
15	A2394C	T774P	More common: Less common:	CGTGGGCTTCACTCAAGAT CGTGGGCTTCACTCAAGAT	236 237
15	G2402C	K776N	More common: Less common:	TCACACTCAAGATCTTCGCTG TCACACTCAAGATCTTCGCTG	238 239
Intron 14	C (+) 16 T	Not applicable	Allele 1: Allele 2:	GCAGCCTCACCCTCTCTCTG GCAGCCTCACCCTCTCTCTG	240 241
17	A2723G	I883M	Allele 1: Allele 2:	AGAAGAGAAATCAGAAAAT AGAAGAGAAATCAGAAAAT	242 243
Intron 17	C (+) 2000 G	Not applicable	Allele 1: Allele 2:	GCGCAGTGCCTGTGTCTCTA GCGCAGTGCCTGTGTCTCTA	244 245
21	T3233G	no change	More common: Less common:	GATCTAAGGTTGTGATTCGG GATCTAAGGTTGTGATTCGG	246 247
Intron 21	G (+) 118 T	Not applicable	Allele 1: Allele 2:	CTCTTCTGTATGACAGAAAGGA CTCTTCTGTATGACAGAAAGGA	248 249
Intron 21	A (+) 563 G	Not applicable	Allele 1: Allele 2:	CATTCTAGGGATCATAGCCAT CATTCTAGGGATCATAGCCAT	250 251
Intron 24	G (+) 321 T	Not applicable	Allele 1: Allele 2:	AAGTACAGTGGGAGAACAGCG AAGTACAGTGGGAGAACAGCG	252 253
Intron 29	A (-) 824 G	Not applicable	Allele 1: Allele 2:	ATTCTCAAAAATAGAAAATGCA ATTCTCAAAAATAGAAAATGCA	254 255
Intron 31	T (+) 30 C	Not applicable	More common: Less common:	GGCCCCGCTTATTATTACT GGCCCCGCTTATTATTACT	256 257
Intron 33	A (+) 732 G	Not applicable	Allele 1: Allele 2:	TGAGAGAAATTAATGAACCCGG TGAGAGAAATTAATGAACCCGG	258 259
Intron 33	C (+) 898 T	Not applicable	Allele 1: Allele 2:	TTTGCTGAAACATCACTGAC TTTGCTGAAACATCACTGAC	260 261
Intron 34	C (+) 234 T	Not applicable	Allele 1: Allele 2:	AACCTCAGTTCCTCATCTGTG AACCTCAGTTCCTCATCTGTG	262 263
34	G4834A	R1587K	More common: Less common:	CTGGACACCAAAATATGTC CTGGACACCAAAATATGTC	264 265
37	C5266G	S1731C	More common: Less common:	TCCTATGTGTCTCCACCAAT TCCTATGTGTCTCCACCAAT	266 267
Intron 43	T (+) 18 C	Not applicable	More common: Less common:	AAGAAGTGGCTGTATTTTGC AAGAAGTGGCTGTATTTTGC	268 269
Intron 43	A (+) 1865 G	Not applicable	Allele 1: Allele 2:	AACCTGATTTGATTGGTATAGCTG AACCTGATTTGATTGGTATAGCTG	270 271
48	C6521T	no change	More common: Less common:	CAGGGTCAACCCGACCTGA CAGGGTCAACCCGACCTGA	272 273
Intron 10	(+) 14 ins. T	Not applicable	More common: Less common:	TCCTCAGGATGGGGACAG TCCTCAGGATGGGGACAG	284 285
Exon 16	G2547A	V825I	More common: Less common:	CCACTTCGATCTCCATG CCACTTCGATCTCCATG	286 287
Polymorphism in an ABC1 BAC contig: This polymorphism is within approximately 200 kb of the ABC1 gene					SEQ ID NO:
	A or G	not applicable	Allele 1: Allele 2:	TTGGGAGGCTAAGGCAGGAGAA TTGGGAGGCTAAGGCAGGAGAA	274 275

Fig. 11



Genomic contig containing ABC1 exon 1:

Underline = putative promotor element

acctcttatagaatgatagaattcctctggaatgattggataacttcatttcaccttgacttttaccttggaggattt  
cttacccttttggcttctcaaatgtgactattaaaatgttgcttttaaaaataggaacacagtttcaggggggagtag  
cagcccatgaccttctgcaaggccccctaactcaaggtagtttccttggaaactgtggtttatggaatgtttcaggagt  
gtgaggaggtataatttaaggctgtccttagcaaggatacccttaaggatagagggccagtagcatctggaggccagaa  
aagttaaactgaggcagtcagattagcttcaggctcaattaagctgatgggtcagcctgggagaaattgcaggatgact  
ctcaatatccccctccacccccacagcagccacgatctgtctgtctttaatcatgggtgcagtgaaactgttctttcca  
gggtgtcttggccttcagtaaccttggttaggcttgctcctgaacgtgggtaccgatccaaagacacatgatcagagaggc  
aattagagaacagaccttttccaaagcaagcatgttctgttgggttagaagtttcatgtcctaattataggacct  
gtgcatctctctggagatgaggcacatgagtcatactgtgattcttctgtctttgtgtcaacatctcatgaataggcaat  
cagagctttggcaccaatgtattttcagttcatatctgtatgttaaataccacctcctgctttgtagtttactggcaa  
gctgttttggatataagacatctagaacactgtaaatataataacatttttattgtctattataacctcaattacgaaaa  
agacatctagaagcaacctcatcaagagagataactgaggccggcatggtagctcacacttgcaatcccattactttgg  
gaggctgaggcaggttagatcacttgaggccaagagtttgaaaccagcctggccaacatgttgaaacctgtctctatta  
aaaaatacaaaaaagtttagctgggcttgggtgggacactgtaatcccagctactccggaggctgaggcaggagaatca  
cttgaacctgggaggcagaggttgagtgagctgagatcacaccactgcactccaacctgggcaccagagtgagattac  
atctaaaaataaaaaaagtaataaaaaagagagataattgatagctgttgttggaatttcaacttccatctcacttc  
tggttaacttttgggaagttgttgaacaaagtggaaatacacgcacatacacacacacatactctcttgtttgttta  
gggttaatagaatagctgtcatataatcactgttttgaagaggagaatttagttgctatctgtacattttgggtatgt  
gaactatttggatagaactctgagaaatgcattcagaacaacaaacaaatcataggagaaatagctaagtgggaaggg  
ccatataagagttgttgaaaaagttatttcttgagaaaccagctctaattgctaggcaagtcacttgctttggggaggc  
ctcagcttctctgtctataagattgcagcaggggtgtagtggaatgagcttcaacattccaagagattttatctact  
aatacgacagtcacaaatggagcatgactttgtggaagcctctcctcttccaccagaggggccaatttctctgtcccagt  
gagatgttgacacttgtatgatccctgcttggagacttccctcttctggaacctgacctgggtcaggcatgagggtga  
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cccatttgttactgcacaagcaaacagcttctggtagctgtacagatacatgcacttcttctcactgtgtttccat  
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tccaagttatgacttgtgaggtatgttaattatgataatagaaggcagtttatttgggtcagatttattgatgtgta  
ttaccacagtaagacttcccccttacaagaatgatgatgttttgacaaatggatacacatgtgtatctaccactgcca  
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tttgggattcatccaggttgtcgcagtaacagtagcttattcctttttaggctgagtaagtgctccagttttattta  
tatatttatttatgaggaggtgtctcactctgtcaccaggtggagtgcggtagcgcgatctcagctcactgcaacct  
ccgctccaggttcaagcaattctcctgctcctgagtagctgggtatcaggcaccacccagccacgccaactaatt  
tttatatttttagtagagatggggtttcaccatgttggccaggtgatctcaaaactcttgacctcaggtgatccgcca  
cctctggctcccaaagtgttaggttacaggcatgagccactgtgcccagccaggtttatttattcaccagttgatg  
gtcttttgcacaactaattgtttccagtttttggctattctgtataaggcttctataaatattcacaatacctaggat  
gggatgactgggtcatataatagtactgtataaccttagcagaaactgtcaaaactattttccaaagtggctcttccatt  
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aatgatattctatttgggttttaatttgcatttctctgcagctaattgatgagtggttctgtctatttgggaagggttta  
atthagcagtcgtgttattctgtagatattaataacttcaaaatcatcagtggtcatttgcagttaaaatttcttaaaa  
aattggccaaagggttccagcagtcacttctgcatgacccaaactgtatgaaacaaggctgaggtgtggagattgtcac  
atthtggcaaggagtgatccacttgggtgactgatgagaccagagagcgtacgcctcgggttgagggtgaggacggg  
cggaagtcgactgcagtgccctgtggccttgggaggctgccagtccttagctaaagctggcagttatgggaaacag



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 gacgtcagctctttgtctctctatctctgaacacccttcccttagagatcccatctctaggatgcatttctctgtagtta  
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 aatgtagcttaacatgtctgtaataaaaatgatcatcttctctgagattcaaagggctataagggacttgggagagaatt  
 tcattcagtttctcctcaaaactagaataatgcttgcactgtctgtaaaagaacaaaagtgtcaaagcatccttttgttca  
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 tatacaactatatgaccatacatatttacaataaagtgcagccaaaattgcaaatcaataccattcaaattaatac  
 cttaaagtgtggtgagggcagctgttgttcaactgaaaccaaattataagttgcatggcagtaaatgctatcatgctgac  
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 aaaaagagagnnnnnnnacacttagaatgagcttccatgtgtgagggcactaactgattaggcattattaactagatttat  
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 a c g t t g t a c a t a g c a g g t t t c a g c a a t g t t a g t c c c t c t t c c c c a c a t c c a t t c a a a t c t g t t c c t t c t c c a a a g g a  
 t g t g t c a a g g a g g a a a t g g a c c t g g c t g g g a a c c c t c a g a a t a c t g g g a t g a t g c t g a g c t t g g c t a c a c c t g t g c t t  
 t g c t t t c a g G C C A G T A C G G A A T T C C C A G G C C T G G T A T T T T C C T T G C A C C A A G T C C T A C T G G T T T G G C G A G G A A A G T G A T  
 G A G A A G A G C C A C C C T G G T T C C A A C C A G A A G A A T G T C A G A A A g t a a g t g c t g t t g a c c t c c t g c t t t t c t t t a a c c t a  
 g t g c t g c t g c c t c t g c t a a c t g t t g g g g g c a a g c g a t g t c t c c t g c c t t t c t a a a a g a c t g t g a a c c a c t c c a g g g g c a  
 g a g a a t c a c a t g c a g t g t c c c t t t c c a a a t c c t c c c a t g c c a t t a t g t c c a a t g c t g t g a c c t a t t g g g a g t t c a c g  
 g t c t c g a t c c c t g a g g g a c a t t t t c t t t g t t g t c t t g g c t t c t a g a a g a g t a t c t t t a c t t g c c c c t c c c a a c a c a c  
 a t t t c a t g g t c t c c t a a c a g c t a g a a g a a g a g g t a a g a c a a g c g t g a t t g t g g a a c c a t a g c c t c g t g c c t g c c t g  
 t g a c a t g g t g a c c t g t g t a t c a g c c t g t g t g g g c t g a g a c c a a g t g g c t a c c a c a g a g c t c a g c c t a t g c t t c a t a a t g t  
 a a t c a t t a c c a g a t c c c t a a t c c t c t c t t g g c t t a a c t g c a g a c a g a g a t g t c c a c a g c t c a t c a a a g g c t c t g c t t  
 c t g g g t t c t t t g t g c t t a g a g t g g c t t c c t a a a t a t t t a a t a g g t c c c t t t c t g c c a g t c t c t t c t g t g c c c a t c c c c t  
 g a t t g c c c t t g g t a a a g t a t g a t g c c c c t t a g t g t a g c a c g c t t g c c t g c t t c c t a a t c a t c t t c t c c t a c c t c c t c  
 t t t a c a c c t a g c t c c t g t t t c a g t c a c c t a g a a a t g c t c a c a g t c g t g g a a t a t g t c a t g t t c t t c c a c a c c t c c a t g c  
 c t t t g t a g g t a c t g t t t g c t c t c a c a g g a a c t t t c t c t a a c t t g c c t a t c t t c t c a a c t c c t c c t t t c t c c a a g  
 a t c t a g t t c c g g a t c c c c t c c c c t g a g c a t c c c t c c t t g g t t c t c a g g t a g t c a g t c a c t c t c t g c c c t g a a c t t c c a t g  
 g c a c g t g a a a g a a a a t c t t t t a t t t t a a a a c a a t t a c a g a c t c a c a a g a a g t a a t a c a a a t t a c a t g a g g g g t t c c c t  
 t a a c c t t t c a t c c a g t t t c c c c a a t g g t a g c a g c a t g t g t a a c t g t a g a a t a g t a t c a a a c c a t g a a a t t g a c a t a g g  
 t a c a a t t c a a a c c t t c t t c a g a t t t c a t a g c t t t a t g t g c g c t c a t t t g t g t g t g t g t g c g t a t t a g t t c t a t g  
 c a a t t t a t c a t g t g t g a a t t c a t g t a a t t a c t a g c t c a g t c a a g c t g c a g a a a t a t c a t t g t c a c a a a g c t c c t t c a  
 t g c t a c c c t t a a t g g c c a c a g c c a c c t c c c t t c t c c t c a g t t c c t g a c a c c t g t c a a c c a c t a a t g c g t t c c t c g t t  
 t t a c a g t t t t a t t a t t t c t a g a a t g t t a c a t a a a t g g a a c c a t a c a g t a g g t a t c c t t t g a t a c t g g c t t t t t t t t t  
 t t t c a c t c a g c a g t a t t c c c t t a g a t c t a t c c a a g t t g t g t g t g t c a a c a g t t c a t t c c t c t c a c t g c t g a g t a g t g t t  
 c c c t g g g a g g g t g t a t c a c a g t t c c a t g g c a t t t t t a g a t g t a t t t t t a a a c a g c t t t c a c a g c t t t c a g c a t c c t a t t t t a a t t  
 g t t c a t c a a g t c c t t t t c c c a a t a g a c t c t g a a t g t c c t t t a t c a t c g t a t t c c c a t c a c c a a c a t c a g t a c c c a a t  
 a g g c c t a a a t a a a c a t t t a t a g c c t c c t g c c t g c c t g a g a a a c c a g g g t g g a c a t g g a g a g a a g g c a c t t c t g a a a g t t  
 c a a g c g c a g t g c s c t g t g t c c t t a c a c t c c a c t c c t c a g t g c t t c t g t g g g t t c a t t t c t g t c t t c t c c t g t c a c a g  
 T C T G C A T G G A G G A G G A A C C C A C C A C T T G A A G C T T G G G C G G T G C C A T T C A G A A C C T G G T A A A A G T C T A C C G A G A T G G G A T G  
 A A G G T G G C T G T C G A T G G C C T G G C A C T G A A T T T T A T G A G G G C C A G A T C A C C T C C T C C T G G G C C A C A A T G G A G C G G G G A A  
 G A C G A C C A C C A T G t a a g a a g a g g g t g t g g t t c c c g c a g a a t c a g c c a c a g g a g g g t t c t g c a g t a g a g t t a g a a t t t a t  
 a c c t t a g g a a a c c a t g c t g a t c c c t g g g c c a a g g g a a g g a g c a c a t g a g g a g t t g c c g a a t g t g a a c a t g t t a t c t a a t c  
 a t g a g t g t c t t t c c a c g t g c t a g t t t g c t a g a t g t t a t t t c t c a g c c t a a a a c a a g c t g g g g c c t c a g a t g a c c t t t c c  
 c a t g t a g t t c a c a g a a t t c g c a g t g g t c t t g g a a c c t g c a g c c a c g a a a g a t a g a t t a c a t a t g t t g g a g g g a g t t g g  
 t a a t t c c c a g g a a c t c t g t c t c t a a g c a g a t g t g a g a a g c a c c t g t g a g a c g a a t c a a g c t g g g c a g c t g g c t t g a t t g  
 c c t t c c c t g c g a c c t a a g g a c c t t a c a g t g g g t a g t a t c a g g a g g g t c a g g g g c t g t a a a g c a c c a g c g t t a g c c t c a  
 g t g g c t t c c a c c a g a t t c c t c a a c c a t t c t a a c c a t t c c a a g g g t a t a t c t t t g g g g g t g a c a t t c t t t t c c t g t t  
 t c t t t t t a a t c t t t t t t a a a c a t a g a a t t a a t a t a t t a t g a g c t t t t c a g a a g a t t t t a a a a g g c a g t c a g a a a t c c  
 t a c t a c c t a a c a c a a a a t t g t t t t a t c t t t g a a a a t a t g t t c t t g t t t g t c a t t t t c c a t g c a t g c g a t g t t a g g c  
 a t a c a a a a t a c a t t t t t t a a a g a a t a c t t t c a t t g c a a a t t g g a a a c t t c g t t t a a a a a t g c t a c a t a a a a t t g g c a  
 t t t c t a a c c c a t a g g c c c a c t t g t a g t t a t t t a c c g a a g c a a a a g g a c a g c t t t g c t t g t g t g g g t c t g g t a g g g t t c a  
 t t a g a a a g g a a t g g g g g c g g t g g g a g g g t g g t g t t c t g t t c t c t g c a g a c t g a a t g g a g c a t c t a g a g t t a a g g g t a  
 g g t c a a c c c t g a c t t c t g t a c t t c t a a a t t t t g t c c t c a g G T C A A T C C T G A C C G G G T T G T T C C C C C G A C C T C G G G C A C  
 C G C C T A C A T C C T G G G A A A G A C A T T C G C T C T G A G A T G A G C A C C A T C C G G C A G A A C C T G G G G G T C T G T C C C C A G C A T A A C G  
 T G C T G T T T G A C A T g t g a g t a c c a g c a g c a c g t t a a g a a t a g g c c t t t t c t g g a t g t g t g t g t c a t g c c a t c a t g g g a g  
 g a g t g g g a c t t a a g c a t t t t a c t t t g c t g t g t t t t g t t t t t c t t t t t t c t t t t t a t t t t t t g a g a t g g a g t c t c g  
 c t c t g t a g c c a g g c t g g a c t g t a g t g g c g c a t c t c g c t c a c t g c a a c c t t g c c t c c c a g g t t c a a g c g a t t c t c c t g  
 c c t c a g c c t c c c a g t a g c t g g g a c t c t a g g c a c a c a c c a c c a t g c c c a g c t a a t t t t t g t g t t t t a g t a g a g a c g g g g  
 t t t c a c c a t g t t g g c c a g g a t g g t c t c a a t g t c t t g a c c t c g t g a t c c g c c a c c t c g g t c t c c c a a g t g c t g g g a a c a  
 c a g g c a t g a g c c a c t g t g t c t g g c c a c a t t t t a c t t t c t t t g a a t a t g g c a g g c t c a c c t c c g t g a a c a c c t t g a g a c c t  
 a g t t g t t c t t t g a t t t t a g c a g a a g t g g g a g g t g a a t g g t t g a g c t g t a g a g g t g a c a t c a g c c c a g c c a g t g g a t g g g g  
 g c t t g g g a a c a t t g c t t c c a t t a t t g t c a t g t g g a g g g c c t t a g c c c a t c c t c t c c c c c g c c a c c c t c c t t a t t  
 g a g g c c t g g a g c a g a c t t c c c a g a c c t g g t a g c t t c a g g g c c c t g g t a t g a t g g a c c t a t a t t t g t g c t t a a g a c a t  
 t t g c t c c c a c t c a g g t t g t c c c a t c a g c c a t a a g g c c c c a g g g a g c c c g t g t g a t g g a g c a g a g a g a c c t g a g c t c t  
 g c a a t c t t g g g c a a g g c t t t c c c t a t g t t t c t t c t a t c t a a a g t g a a c a g c t g g g g c t c a t g t g c t c c c t c c t c a t c  
 t a a a g t g a a c a c a t g g g g c t c a t g t g a g g g t c c t c c c c g c t t t c a g a g c c t g a g g t c c c c t g a g g c t c a g g a a g g c t g c  
 t c c a g g t g a g t g c c g a g c t g a c t t c t t g g t g g a c g t g c t g t g g g g a c a g c c a t t a a a g a c c a c a t c t t g g g g c c c t g a a



Genomic contig containing ABC1 exons 23 to 28:

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Genomic contig containing ABC1 exons 30 and 31:

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Genomic contig containing ABC1 exon 32: \_

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Genomic contig containing ABC1 exons 33 to 36:

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[illegible]

Genomic contig containing ABC1 exons 37 to 41:

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gatt

Genomic contig containing exons 42 through 45:

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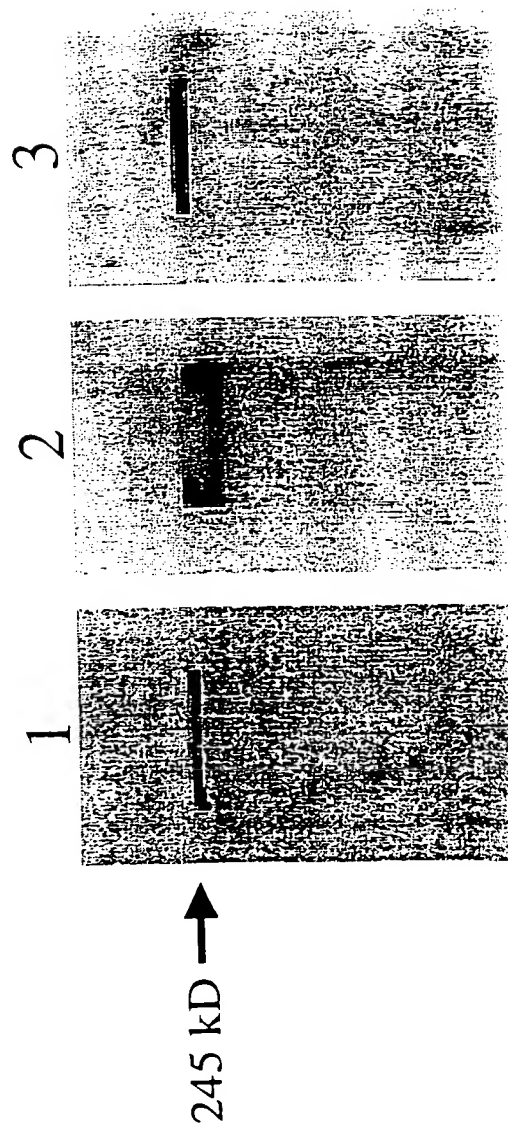


Fig. 13

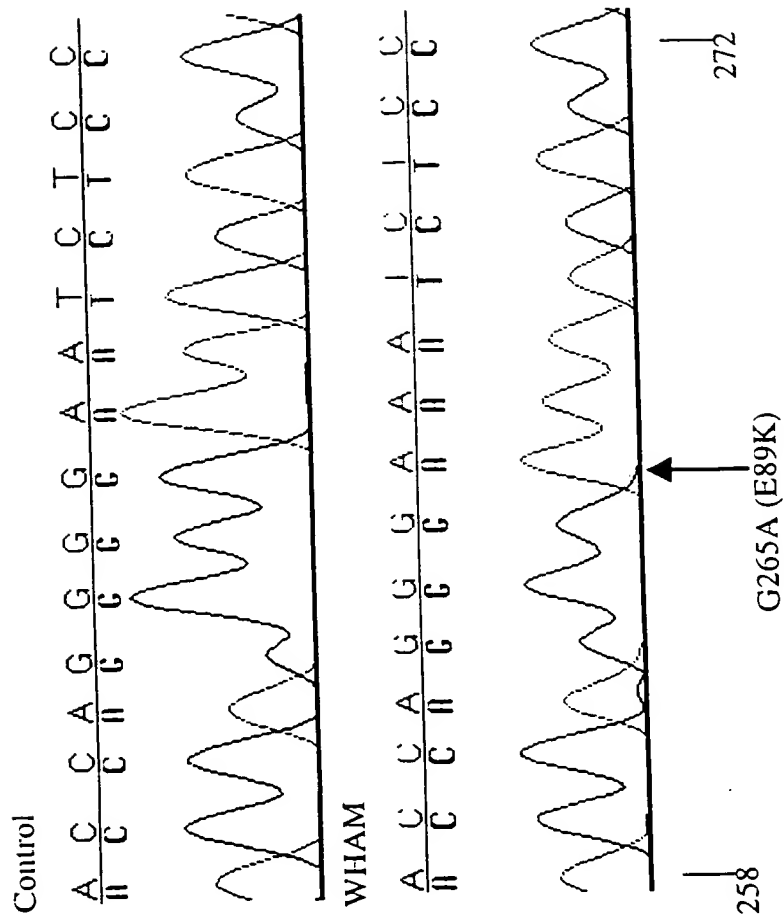


Fig. 14



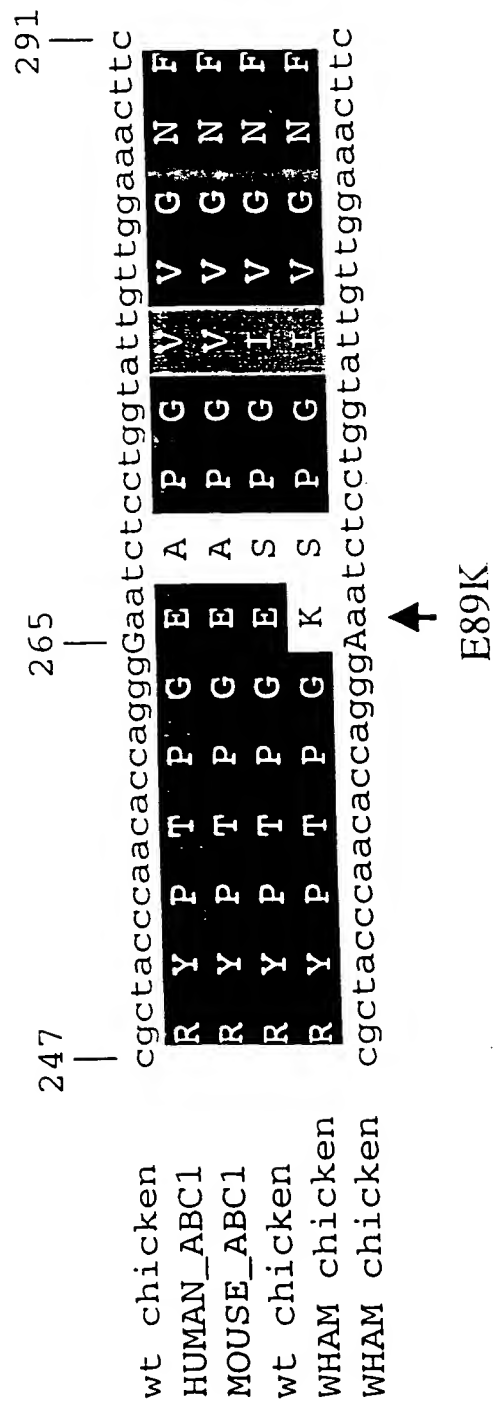


Fig. 15

No.	Name	Location in SEQ ID No. 14	Sequence	Sequence Strand Length
1	PPRE	58-69	AGGTAAAAGTCA	12 Complement
2	PPRE	1997-2009	AGAGTAGAGGGCA	13 Lead
3	PPRE	2150-2161	ATGTCAAAGTTCA	12 Lead
4	PPRE	2156-2169	AGTTCAAAAGGGCA	14 Lead
5	PPRE	4126-4139	AGGCCAGCAGGGCC	14 Complement
6	PPRE	5075-5087	AGGGCAGAAGTGA	13 Lead
7	PPRE	6604-6615	ATGCCAAGGTCA	12 Complement
8	PPRE	6731-6743	GGGGCAAGGTA	13 Complement
9	PPRE	7220-7233	AGGTAATGAGGACA	14 Complement
10	PPRE	7554-7568	GGATCACGAGGTCA	15 Complement
1	SRE	159-166	CAGCCCAT	8 Lead
2	SRE	1133-1140	CAGCTCAC	8 Complement
3	SRE	1145-1152	CACACCAC	8 Lead
4	SRE	1809-1816	CAGCCCTC	8 Complement
5	SRE	1894-1901	CAGCCCAT	8 Lead
6	SRE	2563-2570	CAACCAC	8 Lead
7	SRE	3303-3310	CAGCTCAC	8 Lead
8	SRE	3470-3477	CCGCCAC	8 Lead
9	SRE	4784-4791	CTCCCCAC	8 Complement
10	SRE	4802-4809	CAGCCTAC	8 Complement
11	SRE	4970-4977	CACCTCAC	8 Complement
12	SRE	6487-6494	CAGCCTAC	8 Complement
13	SRE	6565-6572	CACCCAAC	8 Complement
14	SRE	6727-6734	CACCCTAC	8 Lead
15	SRE	7041-7048	CACCCAAC	8 Lead
16	SRE	8059-8066	CAGCCCTC	8 Complement
1	ROR(retinoic acid receptor related)	166-172	AGGGTCA	7 Complement
2	ROR(retinoic acid receptor related)	166-173	AAGGGTCA	8 Complement
3	ROR(retinoic acid receptor related)	363-370	ATGGGTCA	8 Lead
4	ROR(retinoic acid receptor related)	364-370	TGGGTCA	7 Lead
5	ROR(retinoic acid receptor related)	2218-2225	TAGGGTCA	8 Lead
6	ROR(retinoic acid receptor related)	2219-2225	AGGGTCA	7 Lead
7	ROR(retinoic acid receptor related)	3643-3649	TGGGTCA	7 Lead
8	ROR(retinoic acid receptor related)	6604-6610	AAGGTCA	7 Complement
1	SREBP-1 or "E box"	473-479	ACACCTG	7 Complement
2	SREBP-1 or "E box"	536-541	ACACATG	7 Lead
3	SREBP-1 or "E box"	537-543	TCATGTG	7 Complement
4	SREBP-1 or "E box"	555-561	TCATGTG	7 Complement
5	SREBP-1 or "E box"	925-931	ACACTTG	7 Lead
6	SREBP-1 or "E box"	967-973	TCACTTG	7 Lead
7	SREBP-1 or "E box"	968-974	TCAAGTG	7 Complement
8	SREBP-1 or "E box"	1063-1069	ACAGGTG	7 Complement
9	SREBP-1 or "E box"	1104-1110	TCACTTG	7 Lead
10	SREBP-1 or "E box"	1105-1111	TCAAGTG	7 Complement
11	SREBP-1 or "E box"	1561-1567	TCACTTG	7 Lead
12	SREBP-1 or "E box"	1670-1676	TCAAATG	7 Lead
13	SREBP-1 or "E box"	1748-1754	ACACTTG	7 Lead
14	SREBP-1 or "E box"	1749-1755	ACAAGTG	7 Complement
15	SREBP-1 or "E box"	1852-1858	TCATGTG	7 Lead
16	SREBP-1 or "E box"	1853-1859	ACACATG	7 Complement
17	SREBP-1 or "E box"	1899-1905	ACAAATG	7 Complement
18	SREBP-1 or "E box"	2199-2205	ACACGTG	7 Lead
19	SREBP-1 or "E box"	2393-2399	ACAGCTG	7 Complement
20	SREBP-1 or "E box"	2559-27005	ACACCTG	7 Lead
21	SREBP-1 or "E box"	2677-2683	TCACATG	7 Complement
22	SREBP-1 or "E box"	2740-2746	ACAACTG	7 Complement
23	SREBP-1 or "E box"	2969-2975	ACAAATG	7 Lead
24	SREBP-1 or "E box"	2979-2985	ACACATG	7 Lead
25	SREBP-1 or "E box"	2981-2987	ACATGTG	7 Lead
26	SREBP-1 or "E box"	2980-2986	ACATGTG	7 Complement
27	SREBP-1 or "E box"	2982-2988	ACACATG	7 Complement
28	SREBP-1 or "E box"	3461-3467	TCAGGTG	7 Lead
29	SREBP-1 or "E box"	3462-3468	TCACCTG	7 Complement
30	SREBP-1 or "E box"	3547-3553	TCAACTG	7 Complement
31	SREBP-1 or "E box"	3752-3758	ACACATG	7 Lead
32	SREBP-1 or "E box"	4226-4232	TCACCTG	7 Lead
33	SREBP-1 or "E box"	4582-4588	ACACGTG	7 Complement
34	SREBP-1 or "E box"	4588-4594	TCAGTTG	7 Lead
35	SREBP-1 or "E box"	4861-4867	TCAGGTG	7 Lead
36	SREBP-1 or "E box"	4951-4957	ACAAATG	7 Lead
37	SREBP-1 or "E box"	5096-5102	TCAAATG	7 Complement
38	SREBP-1 or "E box"	5912-5918	ACAGTTG	7 Lead
39	SREBP-1 or "E box"	5913-5919	TCAACTG	7 Complement
40	SREBP-1 or "E box"	6245-6251	ACACATG	7 Complement
41	SREBP-1 or "E box"	6288-6294	ACAACTG	7 Complement
42	SREBP-1 or "E box"	6623-6629	TCATTTG	7 Lead
43	SREBP-1 or "E box"	6836-6842	TCACCTG	7 Lead
44	SREBP-1 or "E box"	6837-6843	ACAGGTG	7 Complement
45	SREBP-1 or "E box"	7032-7038	ACAGGTG	7 Complement

Fig. 16

46 SREBP-1 or "E box"  
 47 SREBP-1 or "E box"  
 48 SREBP-1 or "E box"  
 49 SREBP-1 or "E box"  
 50 SREBP-1 or "E box"  
 51 SREBP-1 or "E box"  
 52 SREBP-1 or "E box"  
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 57 SREBP-1 or "E box"  
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 59 SREBP-1 or "E box"  
 60 SREBP-1 or "E box"

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 7240-7246  
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 7654-7660  
 7735-7741  
 7838-7844  
 7880-7886  
 8051-8057  
 8052-8058

TCAGGTG  
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 ACAGTTG  
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7 Lead  
 7 Complement  
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**Fig. 16**